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ABSTRACT

Designed for administrators, teachers, school nurses, and others invclved in health education for kindergarten through adult education, the resource guide provides curriculum ideas for instruction in genetic conditions, heredity, and birth defects. Student learning objectives, content information, learning activities, and evaluation methods are described for subconcepts within the four major goals: to comprehend the role of heredity and environment; to be aware of the most common birth defects and genetic conditions; to acquire basic information about prevention and treatment, and to know where to seek help and further information; and to understand the personal, social, and economic consequences of genetic conditions and birth defects and to consider the ethical implications of medical and genetic advances. (CL)

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ED143162

Genetic Conditions

A Resource Book and Instructional Guide to
Human Heredity and Birth Defects
For Kindergarten Through Adult Education

U S DEPARTMENT OF HEALTH, EDUCATION & WELFARE
NATIONAL INSTITUTE OF EDUCATION

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EC 101416

California State Department of Education
Sacramento, 1977

Cover: Because of genetic factors, no two people have identical fingerprints.

Foreword

Three years ago the Department of Education initiated a special project on instruction about sickle cell anemia. Later the project was expanded to include a wide range of genetic diseases and disorders that warrant particular attention in a comprehensive health education curriculum.

The need for instruction about genetic conditions and birth defects was underscored by the announcement, in the summer of 1976, that a group of American scientists had successfully created an artificial gene. This discovery suggests that entirely new forms of plant and animal life will be created in our lifetime—certainly in the lifetime of our children. It was the latest in a series of discoveries about genetics in the last two decades that have radically changed the thinking of professionals in the fields of science, medicine, and health. As a result of the "new genetics," a great deal of human suffering caused by genetic diseases and genetically related birth defects can now be prevented or alleviated. Students in our schools need to know about these important medical and scientific advances; they need the kind of access to this information that our schools can provide.

This resource book was developed both to inform educators about genetic conditions and birth defects and to assist them in introducing this vitally important aspect of health education into a comprehensive health education curriculum. It presents a wealth of information about human heredity, genetics, and the prevention and treatment of genetic disorders. It offers teachers the option of exploring this fascinating subject in detail or of teaching selected concepts.

Here, for the first time, in a convenient, accessible form, is the information that teachers will need in order to offer instruction about genetic conditions and birth defects. This publication deserves the attention of all educators who are concerned about the health of present and future generations.

Superintendent of Public Instruction

A child with spina bifida, after surgery

Acknowledgments

This publication was developed by the California State Department of Education, Genetic Diseases and Disorders Project, Robert Ryan, Director, in cooperation with the San Diego Regional Center for the Developmentally Disabled.

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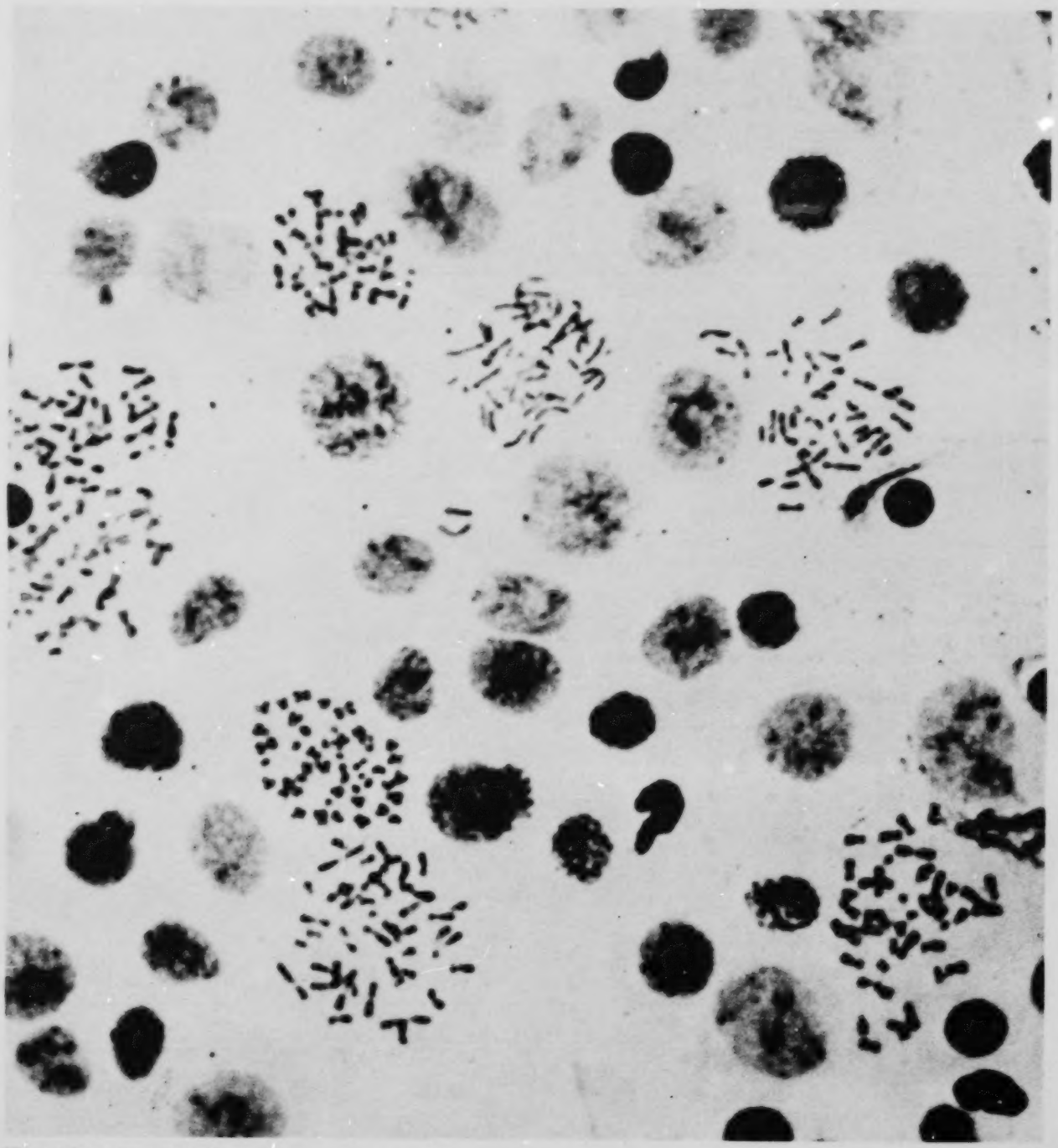

Human chromosomes in culture

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For every child that died in 1906 as the result of a congenital abnormality, or birth defect, six children died from infections or contagious diseases. Today those statistics have reversed: for every child that dies as the result of a contagious disease six die from congenital diseases or disorders. The prevention and treatment of infectious diseases are now commonplace; birth defects, on the other hand, have become a major health problem.

Medical science has actually made it easier for certain kinds of genetic conditions and birth defects to be passed from one generation to another. This is an indirect result not only of advances in obstetrical medicine but of better health care generally. Individuals with what were once considered serious genetic diseases and disorders can now be treated, thus perpetuating those diseases and disorders in their children. Also, the chances of a defective child being born and surviving are greater today than they have ever been before.

It is estimated that as many as 250,000 children are born in the United States every year with major hereditary conditions and disorders that will seriously impair their health. Some birth defects are immediately apparent. Others are not manifested until later in the first few years of life. Still others do not appear until individuals have reached physical maturity or old age. Most serious birth defects, however, can be detected through medical testing during infancy and even before birth.

According to statistics compiled by the National Institutes of Health, 15 million Americans are burdened by some kind of genetic condition or birth defect. The risk of a child being born with such defects is at least three percent for each birth. Genetic diseases and disorders account for approximately one quarter of all in-patient hospital admissions and placements in institutions for the handicapped.

Bambino, sculpture by Andrea Della Robbia

Progress Toward a Solution

Just as medical science has, in a sense, created a new problem by solving old ones, it has also found new ways of treating many birth defects. Even more important, in the last 20 years dramatic advances have been made in detecting congenital defects before birth.

Through prenatal diagnostic techniques prospective parents now can learn a great deal about their unborn child. They can learn with certainty, for example, whether or not the baby is properly positioned for a safe, easy birth; whether it is receiving sufficient oxygen (lack of oxygen can create severe mental retardation, among other birth defects); or whether the position of the placenta may create problems of premature birth or hemorrhaging.

Through even more sophisticated medical and genetic techniques the parents can determine whether the child will be afflicted by numerous diseases or disorders. In many cases the parents can discover important things about the health of their prospective child even before the child has been conceived.

A new medical field has burgeoned as a result of these discoveries: genetic counseling. Using increasingly reliable techniques, genetic counselors can assist prospective parents (usually parents who have already had a child with a birth defect) by predicting numerous conditions that might occur in their unborn children or children they might someday have.

The Role of Education

Efforts to educate the public about the prevention of genetic conditions and birth defects have been limited by a lack of adequate funding and the absence of a well coordinated plan. Thus, it is not surprising that many people have little knowledge about the progress that has been made in diagnosing birth defects. Also, many young people preparing to become parents have never heard of genetic medicine or genetic counseling.

Clearly the schools are a logical starting point for an educational effort regarding genetic conditions and birth defects. Instruction in this area can play a major role in the prevention of the illness and personal tragedy that continue to follow from ignorance. Such instruction is recommended as part of a comprehensive health education curriculum in the *Framework for Health Instruction* adopted by the State Board of Education.

Teachers do not need to become genetic counselors or experts about genetic conditions and birth defects in order to offer instruction about them. Most of the basic factual information teachers will need is contained in this guide. Supplementary materials are readily available from a variety of sources.

What teachers can do is help their students to understand the importance of hereditary factors in personal health, provide them with medical knowledge that will enable them to make well informed decisions about their future children, and motivate them to use medical and counseling resources when necessary.

Human chromosomes

This guide was designed as a resource for administrators, teachers, school nurses, and others who wish to offer instruction about genetic conditions, human heredity, and birth defects. The guide can be used in its entirety as a curriculum planning tool for kindergarten through adult education. Toward this end the guide provides a complete and detailed description of content and learning activities appropriate for the various grade levels.

The guide may also be used selectively. Teachers may wish to offer instruction only about the prevention and treatment of birth defects, for example. If so, the sections of the guide that deal with advanced biology, genetics, and social issues will be primarily useful to the teacher for placing the study of birth defects in a wider context, although they would not necessarily be included as instructional content.

In order to offer instruction about the prevention and treatment of birth defects teachers should know about the many aspects of biology, genetics, and social science that such a study implies. This guide provides the factual information they will need. While teachers may use much of this factual information for reference only, those who wish to explore the numerous related content areas with adults or advanced high school students are given the option of a complete course of study.

The Goals of Instruction About Genetic Conditions and Birth Defects

As outlined in this guide, instruction about genetic conditions and birth defects has four primary student learning goals:

1. To comprehend the ways in which living organisms are shaped both by their heredity and their environment.
2. To be aware of the most common genetic conditions and birth defects and to recognize that certain of these are more likely to occur in specific subpopulations and families.
3. To acquire basic information about the prevention and treatment of genetic conditions and birth defects and to know where to seek help and further information.
4. To understand the personal, social, and economic consequences of genetic conditions and birth defects and to gain insight into the important ethical questions raised by advances in genetic medicine.

In short, students who have received instruction about genetic conditions and birth defects will know that, when confronted with decisions about beginning a family, they will be able to base their decisions on solid information. They will know where to seek additional information and medical help, and they will have some measure of control over their destinies and the destinies of their families.

Children with spina bifida are often paralyzed below the waist.

The Structure and Content of This Guide

The structure of this guide is based on the following four major concepts. Each of these complements one of the four major learning goals.

1. Heredity and environment interact to influence the development of living organisms.

Chemical structure of the DNA molecule, the basis of genes

2. There are more than 2,000 types of genetic conditions and birth defects.
3. Some genetic conditions and birth defects can be prevented, treated, or alleviated.
4. Genetic conditions and birth defects affect individuals, families, and the society.

Each concept is divided into a sequence of subconcepts, progressing logically from one to another in increasing complexity. The guide recommends specific grade levels (primary, intermediate, and secondary/adult education) for each subconcept.

Each subconcept contains several components:

- **Student Learning Objective.** This is a statement of the instructional objective for student learning for the subconcept.

- **Content.** Written primarily for the benefit of the teacher, this section describes the content related to the particular subconcept and its relevance to the main concept, to the general goals of the guide, and occasionally to related curriculum areas. Also included in this section, wherever relevant, is a list of key words with which both the teacher and the students should be familiar when learning about the subconcept.

- **Learning Activities.** Ranging in number from one to three or four, suggested learning activities are offered as a basis for developing classwork and assignments. Many of these activities can be modified for varying grade levels.

- **Suggested Evaluation Activity.** Ways of evaluating student mastery of the instructional objective are suggested for each subconcept. Usually these are stated in the form of topics that will enable the teacher to observe evidence of student learning either through class discussion or in written work.

A Balance of Cognitive and Affective Learning

Many aspects of health instruction—for example, instruction about drug and alcohol abuse—require considerable emphasis on affective learning. Genetic conditions and birth defects comprise a relatively new field, however; thus, factual information must be a primary concern of instruction in this area. One purpose of this guide is to make the necessary information readily available.

Although students should master the facts about genetic conditions and birth defects, inevitably issues arise in such a study concerning students' self-concepts, their attitudes toward others with developmental disabilities, and their attitudes toward the important moral and ethical aspects of genetic medicine and research. Therefore, numerous learning activities are suggested that stress affective learning in addition to cognitive learning. These include provocative questions for student discussions, role-playing exercises, and exercises focusing on feelings and attitudes.

Cognitive learning about genetic conditions and birth defects is, in itself, insufficient for achieving the learning goals recommended in this guide.

Students should be encouraged whenever possible to accept differences that exist among human beings; they should also be motivated to apply their knowledge about genetic conditions and birth defects to their own lives.

Teacher Preparation

Teachers who will provide instruction about genetic conditions and birth defects should become thoroughly familiar with the contents of this guide. Some teachers will already be acquainted with most of the concepts; others may wish to supplement the contents of this guide with readings listed in the reference section. Inservice training may be available from the State Department of Education or community health agencies.

Community Resources

Community agencies can be immensely helpful in supplementing classroom instruction. Frequently they can provide print and audiovisual materials. There may be a genetic counseling service in the community where students can learn about this important medical field by visiting the program or interviewing the staff.

Some genetic conditions and birth defects are more relevant to communities with particular ethnic or racial majorities, and these communities may have local agencies set up to deal with those conditions. In many black communities, for example, organizations exist solely to combat sickle cell anemia.

New Developments in the Field

Although basic principles of heredity and human development have not changed significantly over recent years, there has been a "knowledge explosion" regarding the diagnosis and treatment of specific genetic diseases and disorders.

Research into genetic conditions and birth defects is not only a relatively new field; it is growing rapidly. In the last decade important new discoveries have been made, or public health policies formulated, from one month to another. Therefore, teachers who wish to offer instruction about genetic conditions and birth defects are urged to find ways of staying abreast of current developments. In the reference section several periodicals are listed that can be obtained at little cost or can be found in libraries (see "Selected Readings").

Terminology

One basic aim of this publication is to avoid overly complicated or technical biological and medical terms. Wherever possible, the terminology used in these pages will be easily understood by the average reader.

Nevertheless, oversimplification of highly specialized language has its hazards. Certain technical terms must be learned and fully understood in order to comprehend the contents of this guide. A glossary is provided in the reference section, and key words are also listed in each content section.

Even more important than grasping specific technical terms is having complete clarity about the scope and nature of the guide itself, particularly of what is meant by "genetic conditions" and "birth defects."

- *Genetic conditions.* In the broadest sense any quality that an individual inherits from his or her parents (or earlier generations) is a genetic condition. The color of one's hair and eyes or one's stature and build are inherited characteristics, genetically determined. This guide focuses primarily on genetic diseases and disorders, illnesses or other conditions of malfunction with which an individual is born.

- *Birth defects.* Any disease, disorder, or other condition present at birth that can impair an individual's health, usually in infancy but sometimes not until later in the stages of development, is a birth defect or congenital defect (the terms are synonymous). While a majority of birth defects are genetically related, some are the result of environmental factors—for example, congenital syphilis or numerous handiçaps that can occur when the mother has taken certain drugs or has become infected with rubella during pregnancy. Thus, the term "birth defects" includes both genetic contitions and many other types of diseases and disorders caused by a variety of factors.

The Need for Sensitivity and Tact

Many aspects of instruction about genetic conditions and birth defects may be the source of embarrassment to students who have experienced, either personally or through close relatives, the kinds of medical problems discussed in this guide. Even a relatively minor condition such as poor vision may be a source of embarrassment to a student who is affected. Therefore, teachers should be sensitive and tactful when dealing with content and learning activities that relate to personal or family medical histories.

State Laws Affecting Instruction About Genetic Conditions and Birth Defects

Several learning activities recommended in this guide involve questions about personal aspects of family health. Therefore, teachers should be aware that parents or guardians must be informed of such questions or surveys in writing and that written permission for pupils to participate must be obtained, as provided in Section 10901 of the California Education Code. Section 8506 of the code describes the procedures for offering instruction about human reproduction; although most instruction about genetic conditions and birth defects does not entail specific instruction about human reproduction, teachers should also be aware that section 8506 may apply to some aspects of the learning activities they plan to use.

1

Goal 1: To comprehend the ways in which living organisms are shaped both by their heredity and their environment.

Concept 1: Heredity and environment interact to influence the development of living organisms.

Goal 1: To comprehend the ways in which living organisms are shaped both by their heredity and their environment.

Concept 1: Heredity and environment interact to influence the development of living organisms.

Subconcept	Recommended grade level for instruction*		
	Primary (K–3)	Intermediate (4–6)	Secondary (7–12) & adult education
1.1—Living things produce like organisms.			
1.1.1—Although newborn animals resemble their parents, some features of the parents do not develop in the offspring until later in life.	●	○	○
1.1.2—All people share common traits and characteristics, but no two people are exactly alike.	●	○	○
1.2—In addition to heredity, the environment affects the development of living organisms in many different ways.			
1.2.1—Nutrition is an important factor in the development of living organisms.	●	○	○
1.2.2—It is often difficult to distinguish genetic effects from environmental effects.		●	○
1.2.3—Darwin formulated a theory of evolution and natural selection.		●	○
1.3—Important advances have been made in our knowledge about genetics and human heredity in the last 100 years.			
1.3.1—The invention of the microscope enabled scientists to examine the process of cellular reproduction.		●	○
1.3.2—Mendel discovered the principle of genetically inherited traits and the laws of dominance and recessiveness.			●
1.3.3—Key factors in cellular reproduction are genes, chromosomes, DNA, and the formation of new genetic codes.			●
1.3.4—Mitosis and meiosis are the two basic forms of cellular reproduction; a new genetic code is formed during meiosis.			●

*Note: A solid bullet (●) indicates that this grade level is recommended for teaching this particular subconcept. An open bullet (○) indicates that the subconcept should have been mastered by students at this grade level. Since many students have not received instruction about genetic conditions and birth defects in the earlier grades, instruction may be required at more advanced grade levels for concepts recommended for earlier levels. These recommendations are not intended to be followed rigorously; rather, they are an outline of how a course of study in genetic conditions and birth defects might progress from one grade level to another.

Concept 1: Heredity and environment interact to influence the development of living organisms.

Subconcept 1.1—Living things produce like organisms.

Although newborn animals resemble their parents, some features of the parents do not develop in the offspring until later in life. 1.1.1

Recommended Grade Level: Primary (K–3)

Student Learning Objective

Discuss at least two ways in which a newborn animal resembles its parents and at least two physical qualitities of the parents that have not yet developed in the newborn.

Key Word

Heredity: The inheritance of physical and mental characteristics.

Content

Recognizing that living things produce like organisms is the first step toward understanding heredity. Numerous inherited traits are immediately apparent in newborn animals: body shape, color, prominent features, etc. Other characteristics such as adult size and shape, or length of fur, develop later on.

Learning Activities

1. Have the students cut out magazine pictures of matching pairs of animals, adult and newborn. Play a guessing game: Each student selects an animal picture. Then, without showing their pictures to anyone else, the students guess which animal other students have in their pictures. The object is to acquire the largest number of matched pairs. After the guessing game, the students, either in pairs or in groups, discuss the similarities and differences between the adult and newborn animals.

2. Have the students do a similar matching activity, but instead of using pictures of animals, discuss the similarities and differences between the adult and newborn animals at a zoo.

3. Ask the students to bring in baby pictures. Play a guessing game: Students draw pictures from the pile and name the students to whom they belong. Discuss the physical characteristics that helped to identify the correct students.

Suggested Evaluation Activity

For discussion or writing: Why do newborn animals look the way they look? Why do you look the way you look?

Notes:

Concept 1: Heredity and environment interact to influence the development of living organisms.

Subconcept 1.1—Living things produce like organisms.

1.1.2

All people share common traits and characteristics, but no two people are exactly alike.

Recommended Grade Level: Primary (K–3)

Student Learning Objective

Identify at least two ways in which students in the class resembl each other physically and two ways in which they are different.

Key Word

Trait: A distinguishing quality or characteristic.

Content

All human beings are distinguished by certain universal physical characteristics: arms, legs, hair, body shape, etc. There is so much variation in these characteristics from one person to another, however, that no two people are exactly alike. Physical differences among racial and ethnic groups are one extreme of the spectrum of human variation; less dramatic differences include the shapes of faces, noses, and limbs.

Learning Activities

1. Examine or make drawings of different types of physical features: Roman, ski-jump, or pug noses; round or slanted eyes; different shapes of mouths. Have the students make a composite drawing of a face, choosing from the various types of features.

2. The class divides into pairs. Each pair lists five shared physical qualities and five differences. Selected pairs explain their findings to the rest of the class.

3. Fingerprinting. Using a rubber stamp pad, each student makes a copy of his or her fingerprint. Then the class members compare fingerprints.

Suggested Evaluation Activity

For discussion or writing: In what ways are all people similar? Different?

Notes:

Concept 1: Heredity and environment interact to influence the development of living organisms.

Subconcept 1.2—In addition to heredity, the environment affects the development of living organisms in many different ways.

Nutrition is an important factor in the development of living organisms. 1.2.1

Recommended Grade Level: Primary (K–3)

Student Learning Objective

Describe how the physical development of a particular living organism can be affected by poor nutrition.

Key Words

Environment: The conditions or influences present in the world an individual inhabits.

Nutrition: Any form of nourishment, mainly food.

Content

Next to heredity nutrition is the most important influence on the development of an organism. Whatever its inherited traits, no organism can grow into a healthy adult without proper and adequate nutrition. In human beings inadequate nutrition can produce numerous diseases and disorders such as rickets, brain damage, and beriberi. (Note: This concept does not deal with nutritional factors affecting prenatal development. See 2.3.1.)

Learning Activities

1. In the classroom plant seeds from the same packet in separate containers. Each container receives a different kind of nutrition—more or less light, water, plant food, etc. (For simplicity, limit each container to one variable only.) Note the development of the seeds during a period of several weeks.

2. To illustrate the need for minerals in one's diet for the growth of bones: Soak a small uncooked bone in vinegar for three days. The mineral matter will dissolve, and the bone will lose its strength and firmness so that it can be bent easily.

3. Find pictures of various foods that humans need for healthy growth and development. Use the pictures to make a wall collage.

—Continued

Notes:

Concept 1: Heredity and environment interact to influence the development of living organisms.

Subconcept 1.2—In addition to heredity, the environment affects the development of living organisms in many different ways.

1.2.1 (continued)

Nutrition is an important factor in the development of living organisms.

4. For secondary or adult education students: Discuss diseases that can result from deficiencies in vitamins, minerals, and protein. Many of these diseases are more common in foreign countries; try to find pictures of people who have the diseases. Discuss which foods contain the necessary nutritional qualities.

Examples:	*Deficiency*	*Disease*
	Protein	Kwashiorkor, Edema
	Calcium/Vitamin D	Rickets
	Vitamin A	Night blindness
	Vitamin B_1	Beriberi
	Niacin	Pellagra
	Vitamin C	Scurvy

Suggested Evaluation Activity

For discussion or writing: What elements of nutrition make a person grow healthy and strong?

1.2.2

It is often difficult to distinguish genetic effects from environmental effects.

Recommended Grade Level: Intermediate (4–6)

Student Learning Objective

Cite at least two examples in which it may be difficult to distinguish genetic effects and environmental effects on the development of an organism.

Twins have been the subject of extensive research to determine the effects of heredity and environment.

Content

The relative influences of heredity and environment, nature and nurture, have been debated by philosophers and scientists for centuries. Discoveries in the field of genetics have done little to dull the debate.

Although purely genetic causes have been determined for 20 percent of known birth defects, the majority of them are probably caused by an interaction of hereditary and environmental factors.

In recent years Richard Herrnstein, Arthur Jensen, William Shockley, and several other educator/researchers have argued that intelligence and academic achievement derive primarily from genetic factors. This idea has been strongly opposed by those who favor the concept of equal op-

—Continued

It is often difficult to distinguish genetic effects from environmental effects.

1.2.2
(continued)

portunity and who believe that schools can produce it by correcting the disadvantages of poverty.

No matter how heated the nature/nurture debate has been, no conclusive proof exists that either heredity or environment is the sole determinant of human behavior, inteliigence, physical characteristics, or the majority of congenital defects.

Learning Activities

1. On the board draw a continuum like the one below:

Determined by . . .

Heredity — Environment

Referring to the following list, for each item select a student to come up to the board and place a mark where he or she thinks the item belongs on the continuum. Ask the student to explain this choice; then open the topic to class discussion.

Items:

The color of a person's eyes
The color of a person's hair
A horse's dappled coat
A student's popularity among his or her friends
A pleasant personality
Doing well in school
Getting into trouble with adults
Being a good runner
Right-handedness or left-handedness
The ability to roll one's tongue

2. Class exercise: Identify physical traits or traits of character that you possess that resemble those of your parents, grandparents, aunts, uncles, or cousins. Make a list of traits. Put an "H" after those you think are due to heredity and an "E" after those you think are the result of environmental factors. Can you be positive about this? For advanced students: Discuss how an experiment might be set up to determine whether such traits are determined by heredity or environment.

3. For secondary or aduit education students: Have students read about, and report to the class on, studies of separated identical twins. Resource: Ashley Montague, *Human Heredity*, chapters 7 and 8 (see "Selected Readings" in the reference section).

Suggested Evaluation Activity

For discussion or writing: When can you be sure whether your physical qualities are the result of either heredity or environment? Give examples.

Concept 1: Heredity and environment interact to influence the development of living organisms.

Subconcept 1.2—In addition to heredity, the environment affects the development of living organisms in many different ways.

1.2.3

Darwin formulated a theory of evolution and natural selection.

Recommended Grade Level: Intermediate (4–6)

Student Learning Objective

Discuss the importance of Charles Darwin's discoveries, and briefly summarize the theories of evolution and natural selection.

Key Words

Evolution: Gradual change from one form to a new, or different, form.

Species: Groups of living organisms that interbreed.

Content

In the mid-nineteenth century the English biologist Charles Darwin touched off a controversy that has continued into the present. Darwin challenged the prevailing notion of the creation of all living things on earth. Until Darwin, it was commonly believed in the West that all living things were created at one moment in time, about 6,000 years ago, independent of each other. Basing his theories on a lifetime of biological research and several years of travel through the seas of the world aboard the H.M.S. *Beagle*, Darwin advanced a new theory with two basic components. Greatly simplified, these are:

1. *The theory of evolution.* All living things evolved very slowly from a common ancestry, going back millions of years; therefore, all living things are related to each other.
2. *The theory of natural selection.* Since all living things undergo a process of continuing evolution, a constant struggle for survival occurs in which some species live while others perish. Darwin used the phrase "the survival of the fittest" to describe this process.

Darwin's theories opened the way for more advanced investigation into the process of heredity.

—Continued

FINCHES.

Darwin's observation of finches in the Galapagos Islands inspired his theory of evolution.

Notes:

Concept 1: Heredity and environment interact to influence the development of living organisms.

Subconcept 1.2—In addition to heredity, the environment affects the development of living organisms in many different ways.

Darwin formulated a theory of evolution and natural selection. **1.2.3** (continued)

Note: In accordance with policies of the California State Board of Education it is recommended that instruction about the theory of evolution be balanced by instruction about other theories of the origin of living things. For further information refer to: *Social Sciences Education Framework for California Public Schools*, page 38; also, *Science Framework for California Public Schools* and its 1974 addendum.

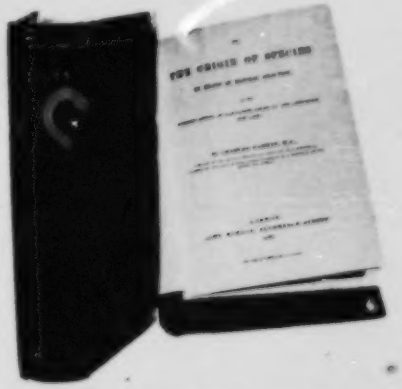
Learning Activities

1. Have the class invent or explore alternative theories about how living things came into being.

2. Divide the class into groups. Each group is assigned a particular living organism. Examples: birds, rodents, wolves, deer, whales, or tribes of people such as the Australian aborigine. Then each group designs a sequence of possible alternative future evolutions for this organism, applying the theories of evolution and natural selection.

Suggested Evaluation Activity

Discuss: Why is Darwin's theory of evolution important to our knowledge about human heredity? In your discussion, demonstrate that you understand what is meant by "evolution" and "natural selection."



A first edition of Darwin's *The Origin of Species*

Notes:

Concept 1: Heredity and environment interact to influence the development of living organisms.

Subconcept 1.3—Important advances have been made in our knowledge about genetics and human heredity in the last 100 years.

1.3.1

The invention of the microscope enabled scientists to examine the process of cellular reproduction.

Recommended Grade Level: Intermediate (4–6)

Student Learning Objective

Explain how the invention of the microscope contributed to the modern science of genetics.

Key Words

Cell: The living active microscopic unit of all plants and animals, consisting of many specialized parts.
Chromosomes: Microscopic threadlike bodies in the nuclei of cells.
Cytology: The study of cells.
Genes: The units in chromosomes that determine hereditary characteristics.
Nucleus: The central part of a cell.
Sperm: Male germ (sex) cells that unite with female ova (egg) cells to produce a new organism.

Content

Although it is now known that the reproduction of cells holds the key to heredity in all living things, cells were not seen by humans until the development of the first modern microscopes by Antoni van Leeuwenhoek, a Dutchman, in the late 1600s. Leeuwenhoek spent many years examining the cells of animals and plants and was the first to observe the human sperm cell.

Leeuwenhoek's experiments opened the way for far more sophisticated microscope technology and experimentation that led to a surge of discoveries about reproduction in the nineteenth and twentieth centuries. In fact, Leeuwenhoek's work made possible a new field of biological science: cytology, the study of cells. In chronological order, some of the most important cytological discoveries after Leeuwenhoek included:

1831: First observation is made of cell nuclei.

1839: Biologists M. J. Schleiden and Theodor Schwann prove that cells are the basic units of all organisms.

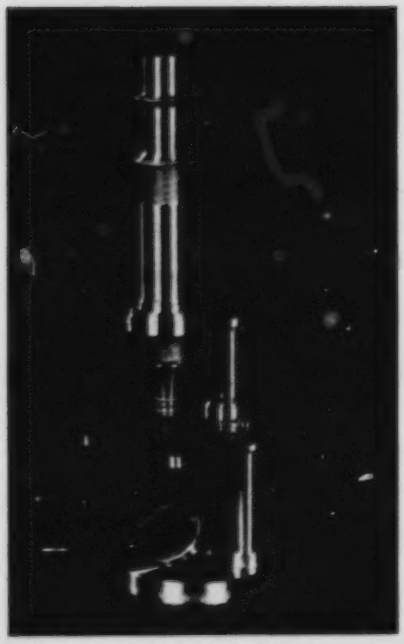
1875–76: Division of nuclei is observed.

1880: Biologist Walter Flemming applies dye to the nucleus of cells; the dye makes visible tiny threadlike bodies that came to be known as chromosomes ("colored bodies").

1940s: Multiple discoveries and new theories indicating that the chromosomes contain the basic elements of heredity, the genes.

1950s: The discovery of the chemical structure of deoxyribonucleic acid (DNA), the chemical of which genes are made.

—Continued



A microscope of the mid-nineteenth century

Concept 1: Heredity and environment interact to influence the development of living organisms.

Subconcept 1.3—Important advances have been made in our knowledge about genetics and human heredity in the last 100 years.

The invention of the microscope enabled scientists to examine the process of cellular reproduction.

1.3.1
(continued)

Learning Activities

1. Select two or three students to do research on the discoveries described above and have them report the details of their findings to the class.
2. Observe different types of cells through microscopes.
3. Make drawings of different types of cells, tracing pictures from books in the school library.

Suggested Evaluation Activity

Have the students write a paragraph on the topic "Without Microscopes...."

Notes:

A modern microscope

Concept 1: Heredity and environment interact to influence the development of living organisms.

Subconcept 1.3—Important advances have been made in our knowledge about genetics and human heredity in the last 100 years.

1.3.2

Mendel discovered the principle of genetically inherited traits and the laws of dominance and recessiveness.

Recommended Grade Level: Secondary (7–12) and adult education

Student Learning Objective

Discuss Mendel's contribution to the modern science of genetics.

Gregor Mendel

Key Word

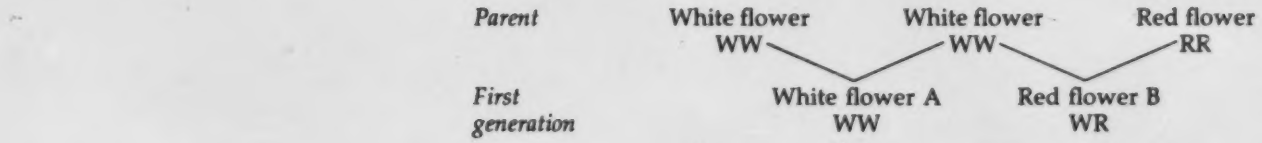
Genetics: The scientific study of heredity.

Content

Charles Darwin had no acquaintance with the pioneering work of the Austrian botanist Gregor Mendel, although both men achieved contributions to biological science at about the same time in history. Unlike the writings of Darwin, Mendel's theory of heredity created scarcely a stir when it was published in 1866. It was not until more than 30 years later that Mendel's work was re-discovered and he became widely recognized as the father of modern genetics.

Mendel's scientific method was based on the observation of distinct inherited traits in the pea plants he studied in his monastery garden (the traits included size, color, and shape). Thus, the Mendelian principles of heredity were derived solely from deductive reasoning.

Mendel determined that each isolated trait was passed on from one generation to another by a single pair of hereditary units, one unit being derived from each parent. Today we call these units genes. The following diagram illustrates hereditary patterns in the offspring of a hypothetical flowered plant (in genetics, particular traits are identified by pairs of letters, as in the diagram):



The Mendelian "laws" of heredity:

1. *The law of dominance.* When organisms with differing traits of a pair are crossed, only one trait, the dominant trait, of the pair appears in the first generation. The white- and red-flowered parents in the example above are pure for those traits. Red is dominant; therefore, the first generation is red. The trait that does not appear is a recessive trait.

2. *The law of segregation.* The characteristic that was hidden (recessive) in the first generation is not lost, since it reappears in later generations. Flower B will pass the recessive white-flower trait to a future generation.

3. *The law of independent assortment.* Every characteristic is inherited independently of every other characteristic. According to this law, new

—Continued

Mendel discovered the principle of genetically inherited traits and the laws of dominance and recessiveness.

1.3.2
(continued)

combinations of traits are possible in succeeding generations; this contradicts the common belief prior to Mendel's discoveries that distinct traits, like paints mixed in a can, disappeared in the mixing process.

Learning Activities

1. Conduct classroom experiments that imitate Mendel's experiments, breeding and cross-breeding simple varieties of plants or flowers. Resource: the *World Book Encyclopedia* article on "Heredity" offers excellent instructions for such an experiment, using fruit flies.

2. Visit a farm or orchard in the area and inquire about the ways in which improved varieties of fruit and vegetables are developed.

3. Invite an agriculture extension agent from your county or a nearby county to speak to the class about improved seeds or plants used in the area. Ask the agent to describe the research that produced the seeds.

4. Survey the class for dominant and recessive traits. Try to guess from the numbers of such traits among the class members and their families which traits are dominant and which are recessive.

Dominant	*Recessive*
Ability to taste phenylthiocarbamide (PTC)*	Inability to taste PTC
Ability to roll one's tongue	Inability to roll one's tongue
Brown eyes	Blue eyes
Curly hair	Straight hair
Free earlobes	Attached earlobes
Long eyelashes	Short eyelashes

Suggested Evaluation Activity

For discussion or writing: Describe the Mendelian laws of inheritance. Why is Mendel often called the father of modern heredity?

*Taste papers available from: American Genetics Association
1507 M Street, N.W.
Washington, D.C. 20005

Notes:

Cattle-breeders draw on principles of heredity to produce animals like this champion steer.

Concept 1: Heredity and environment interact to influence the development of living organisms.

Subconcept 1.3—Important advances have been made in our knowledge about genetics and human heredity in the last 100 years.

1.3.3

Key factors in cellular reproduction are genes, chromosomes, DNA, and the formation of new genetic codes.

Recommended Grade Level: Secondary (7–12) and adult education

Student Learning Objective

Discuss the basic elements of cellular reproduction.

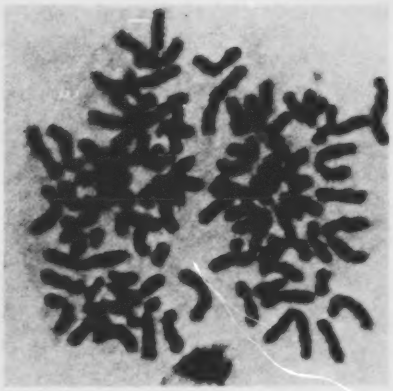
Key Word

Cytogenetics: The study of the genetic effects of cellular reproduction, focusing primarily on the chromosomes.

Content

In the half-century after Mendel's work was fully recognized (1900–1950) the new field of cytogenetics grew rapidly. Important discoveries were made (see 1.3.1) that led to a complete picture of how cellular reproduction transmits genetic information. New discoveries in cytogenetics continue to be made. The basic components of cellular reproduction are:

- *Chromosomes.* These thin threads, located in the nucleus of every cell in the body of an organism, contain all the genetic information needed to reproduce that organism. Some organisms have as few as four chromosomes in their cells. Human beings have 46 (23 pairs, half of these being contributed by the sperm cell and the other half by the egg cell). One pair is called sex chromosomes; these determine the sex of the offspring. The remaining 44 chromosomes are called autosomes. A single human chromosome is extremely small—about two thousandths of an inch, or one eightieth of a millimeter, long—and can be seen only under a high-powered microscope.



Human chromosomes

- *Genes.* No one has ever actually seen this ultramicroscopic unit of heredity. The name, derived from the Greek *gen* (kind), came to be used by geneticists to describe those parts of the chromosomes that contain the information to produce hereditary traits. Genes are often compared to individual beads strung along the chromosomes. Anywhere from thousands to millions of genes may exist in a single chromosome.

- *DNA.* Deoxyribonucleic acid, the structure of which was discovered in the early 1950s, is the chemical essence of genes. DNA is like a ladder; the two sides are connected by numerous combinations of rungs made up of four chemical compounds. In the process of cellular reproduction the two sides of the ladder come apart and duplicate themselves to produce new chromosomes.

- *The genetic code.* This term is used to describe the process of chemical change and recombination that occurs during cell division. The four basic chemical compounds of DNA and the 20 amino acids that make up all proteins are the components of the genetic code, which is analagous to the alphabet that makes up a language. The potential of the various chemicals in cells to form new combinations is far greater than that of the alphabet, however; the virtually infinite number of new combinations accounts for the diversity of living organisms.

—Continued

Concept 1: Heredity and environment interact to influence the development of living organisms.

Subconcept 1.3—Important advances have been made in our knowledge about genetics and human heredity in the last 100 years.

Key factors in cellular reproduction are genes, chromosomes, DNA, and the formation of new genetic codes.

1.3.3
(continued)

Learning Activities

1. In order to grasp the infinite capacity of the various chemicals in human cells to form genetic codes have the students figure out how many possible combinations there could be:
   - Of five different letters
   - Of five different flavors of ice cream combined with five different toppings
2. Have students review such newspapers and magazines as *Time*, *Newsweek*, the *Reader's Digest*, or *Science Year* for articles relating to research on genetics. Prepare a bulletin board of these and discuss them in class.
3. Independent learning activity for advanced students: Read and discuss James D. Watson's *The Double Helix*, an account of how the structure of DNA was discovered (see "Selected Readings" in the reference section).

Suggested Evaluation Activity

For discussion or writing: Explain the importance to genetics of chromosomes, DNA, and the genetic code.

Notes:

Concept 1: Heredity and environment interact to influence the development of living organisms.

Subconcept 1.3—Important advances have been made in our knowledge about genetics and human heredity in the last 100 years.

1.3.4

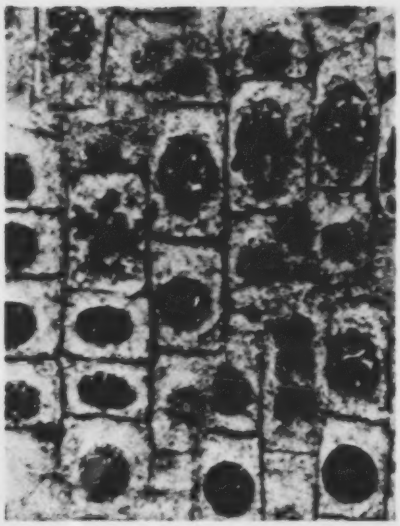
Mitosis and meiosis are the two basic forms of cellular reproduction; a new genetic code is formed during meiosis.

Recommended Grade Level: Secondary (7–12) and adult education

Student Learning Objective

Distinguish between mitosis and meiosis, and discuss the importance of meiosis in creating human diversity.

Content



Onion root cells in varying stages of mitosis

The observation of mitosis and meiosis, the two basic forms of cellular reproduction, occurred in the 1880s. This enabled a more complete knowledge of human heredity than had ever been possible before and confirmed biological phenomena that Mendel guessed at but never saw.

Mitosis is the reproduction of a single cell. This process begins in the nucleus with the division of the chromosomes. After the chromosomes have produced a duplicate of themselves a new nucleus forms, and finally an exact duplicate of the first cell is created. Cells continually divide (and later die); there may be as many as one hundred trillion cells in an adult human being.

Meiosis, the division of sex (germ) cells, is a considerably more complex process than mitosis. Both male sperm cells and female egg cells undergo meiosis.

Unlike mitosis, meiosis occurs in two phases. The first division produces two cells containing double-stranded chromosomes; the second produces four cells containing single-stranded chromosomes.

Learning Activities

1. Observe the cell division of amoeba under a microscope. Prepared slides are available in many school science centers.

2. Show the film *Human Heredity*, available in many school resource centers or from:

I.C. Brown Trust Co.
3170 S.W. 87th Avenue
Portland, Oregon 97225

Suggested Evaluation Activity

For discussion or writing: Describe two important differences between mitosis and meiosis.

Notes:

2

Goal 2: To be aware of the most common genetic conditions and birth defects and to recognize that certain of these are more likely to occur in specific subpopulations and families.

Concept 2: There are more than 2,000 types of genetic conditions and birth defects.

Goal 2: To be aware of the most common genetic conditions and birth defects and to recognize that certain of these are more likely to occur in specific subpopulations and families.

Concept 2: There are more than 2,000 types of genetic conditions and birth defects.

Subconcept	Recommended grade level for instruction*		
	Primary (K–3)	Intermediate (4–6)	Secondary (7–12) & adult education
2.1—Although genetic conditions and birth defects are common health problems, certain conditions appear more frequently than others.			
2.1.1—Contrary to popular belief, genetic conditions and birth defects are common health problems.		●	○
2.1.2—Among more than 2,000 known genetic diseases and conditions only a relative few are commonly found in the general population.		●	○
2.1.3—Certain genetic conditions are associated with particular ethnic or racial groups.		●	○
2.2—Some defects are manifested at birth; others do not appear until later in life.			
2.2.1—Many serious congenital diseases and disorders appear during the first two years of life.		●	○
2.2.2—Some congenital diseases and disorders occur in later stages of development.		●	○
2.3—Birth defects can be the result of a variety of factors.			
2.3.1—Factors affecting the mother's health during pregnancy can cause birth defects (poor nutrition, drug or alcohol use, contagious diseases, etc.).		●	○
2.3.2—Although premature birth is not itself a defect, it can affect the child's later development.			●
2.3.3—Birth defects are frequently the result of genetic factors.			●
2.4—Genetic conditions are transmitted in several different ways.			
2.4.1—Some genetic conditions are the result of chromosome abnormalities.			●
2.4.2—Some genetic conditions are the result of one of three single-gene defects: dominant inheritance, recessive inheritance, or X-linked inheritance.			●
2.4.3—Some genetic conditions are the result of multifactorial inheritance.			●

*Note: A solid bullet (●) indicates that this grade level is recommended for teaching this particular subconcept. An open bullet (○) indicates that the subconcept should have been mastered by students at this grade level. Since many students have not received instruction about genetic conditions and birth defects in the earlier grades, instruction may be required at more advanced grade levels for concepts recommended for earlier levels. These recommendations are not intended to be followed rigorously; rather, they are an outline of how a course of study in genetic conditions and birth defects might progress from one grade level to another.

Concept 2: There are more than 2,000 types of genetic conditions and birth defects.

Subconcept 2.1—Although genetic conditons and birth defects are common health problems, certain conditions appear more frequently than others.

Contrary to popular belief, genetic conditions and birth defects are common health problems. 2.1.1

Recommended Grade Level: Intermediate (4–6)

Student Learning Objective

Explain why genetic conditions are more common than is usually believed.

Content

The notion that genetic conditions and birth defects are "someone else's problem" and exceedingly rare is a common myth. Considering that research into genetic conditions and birth defects is still a relatively young field, what is now known about the extent and seriousness of the problem may merely be the tip of an iceberg. Approximately 15 million Americans are affected by birth defects; of these, 12 million are the victims of diseases involving defects in the genes or chromosomes.

A 42-day-old human embryo

Other statistics*:

- Of all spontaneous abortions (miscarriages), 36 percent are caused by gross chromosome defects (more than 100,000 per year).
- At least 40 percent of all infant mortality is the result of genetic factors.
- Genetic defects are present in nearly five percent of all live births.
- About one third of all patients admitted to hospital pediatric wards are there for genetic reasons.
- Everyone carries between five and eight recessive genes for genetic disorders; while the carrier may not be affected, disorders may occur in future generations.

In addition to these statistics, geneticists and physicians are beginning to find significant links between genetic factors and a wide variety of diseases and disorders that were not previously considered to be genetically related. These include heart disease, certain forms of arthritis, cancer, and some forms of mental illness.

The chances of higher incidences of genetic disease are increasing. Treatment for many genetic conditions has enabled individuals who might once have died or suffered severe handicaps to lead relatively normal lives, have families, and pass genetic defects on to future generations.

—Continued

*Source: *What Are the Facts About Genetic Disease?* Booklet published by National Institutes of Health (see "Instructional Materials for Classroom Use" in the reference section).

Concept 2: There are more than 2,000 types of genetic conditions and birth defects.

Subconcept 2.1—Although genetic conditions and birth defects are common health problems, certain conditions appear more frequently than others.

2.1.1 (continued)

Contrary to popular belief, genetic conditions and birth defects are common health problems.

Learning Activities

1. For class discussion: How many students know someone who was born with any kind of physical problem or abnormality, however minor? Include such examples as poor vision and birthmarks.

Suggested Evaluation Activity

For class discussion or writing: Cite evidence that genetic conditions and birth defects are not as rare as is usually believed.

2.1.2

Among more than 2,000 known genetic diseases and conditions only a relative few are commonly found in the general population.

Baby with cleft lip, before surgery

Recommended Grade Level: Intermediate (4–6)

Student Learning Objective

Identify at least three of the most common genetic diseases and conditions.

Content

Approximately 2,000 genetic diseases and conditions are known to exist, and as many as 100 additional genetically related diseases and disorders are discovered every year. The following list of commonly found diseases and disorders refers only to conditions that are known to be genetically related. Other serious defects such as prematurity or conditions caused by contagious diseases are not included.

Selected high-incidence genetic diseases and disorders

Condition	*Incidence*
Cleft lip/palate	1 in 1,000
Club foot	1 in 1,000
Cystic fibrosis	1 in 2,000
Diabetes	1 in 2,000
Down's syndrome	1 in 1,000
Hemophilia	1 in 10,000
Muscular dystrophy	1 in 20,000
Phenylketonuria (PKU)	1 in 10–12,000
Rh incompatibility	1 in 100
Sickle cell anemia	1 in 500
Tay-Sachs disease	1 in 3,600
Thalassemia (Cooley's anemia)	Not available

—Continued

Concept 2: There are more than 2,000 types of genetic conditions and birth defects.

Subconcept 2.1—Although genetic conditions and birth defects are common health problems, certain conditions appear more frequently than others.

Among more than 2,000 known genetic diseases and conditions only a relative few are commonly found in the general population.

2.1.2
(continued)

Other congenital problems that may or may not be genetically related occur in even greater numbers. These include hearing and visual impairments, heart and circulatory defects (30–40,000 babies every year), and prematurity (nearly 250,000 babies every year).

Learning Activities

1. Class exercise: Ask the students to note, on a scale of 1 to 10, how much difficulty adjusting to everyday life experiences a person would have if affected by each of the conditions listed above.

2. Discuss: If you could plan a telethon to benefit the victims of a particular congenital disorder, which one would you select? What ideas do you have for skits, slogans, or other information to be presented during the show?

Suggested Evaluation Activity

For discussion or writing: Arrange the following genetic conditions in order, beginning with those of highest incidence and ending with those of lowest incidence:

Phenylketonuria (PKU)	Muscular dystrophy
Cleft lip/palate	Cystic fibrosis
Hemophilia	Tay-Sachs disease
Rh incompatibility	Diabetes

Notes:

Baby on opposite page, after surgery to correct cleft lip

Concept 2: There are more than 2,000 types of genetic conditions and birth defects.

Subconcept 2.1—Although genetic conditions and birth defects are common health problems, certain conditions appear more frequently than others.

2.1.3

Certain genetic conditions are associated with particular ethnic or racial groups.

Recommended Grade Level: Intermediate (4–6)

Student Learning Objective

Identify at least two genetic conditions that predominate in different ethnic or racial groups.

Content

Among the genetic diseases and disorders that are more commonly found in the general population some occur almost entirely within a particular ethnic or racial group. The geographical origins of the ethnic or racial group can also be an important factor.

Condition	*Ethnicities primarily affected*
Cleft lip/palate	All (more common in Orientals)
Cystic fibrosis	Caucasians
Phenylketonuria (PKU)	Caucasians and Orientals
Sickle cell anemia	Blacks and Mediterraneans
Tay-Sachs disease	Ashkenazi Jews (origins in central and eastern Europe)
Thalassemia	Mediterraneans

Learning Activities

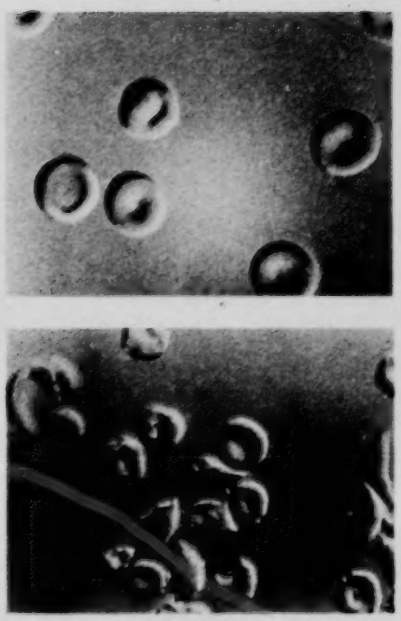
1. Have a student volunteer report to the class on the distinction between sickle cell anemia and sickle cell trait.

2. Discuss the advantages and disadvantages of screening programs to detect genetic diseases in different ethnic and racial groups.

Suggested Evaluation Activity

For discussion or writing: Which genetic conditions are most likely to occur among the following ethnic or racial groups?

Caucasians	Mediterraneans
Blacks	Orientals
Jews	



Above, normal blood cells; below, sickled blood cells

Notes:

Concept 2: There are more than 2,000 types of genetic conditions and birth defects.

Subconcept 2.2—Some defects are manifested at birth; others do not appear until later in life.

Many serious congenital diseases and disorders appear during the first two years of life. 2.2.1

Recommended Grade Level: Intermediate (4–6)

Student Learning Objective

Describe at least two congenital diseases or disorders that may appear during the first two years of life.

Key Word

Psychomotor: Of or relating to motor action directly proceeding from mental activity.

Content

Some congenital defects are immediately observable at birth. These include physical malformations such as cleft lip/palate, club foot, and polydactyly (additional fingers and toes). While others may be present at birth, testing is required to confirm their presence. A newborn child may have an unusually large head, for example, a possible symptom of hydrocephalus, a condition of abnormal absorption of liquids within the central nervous system that can be confirmed through X-ray tests; phenylketonuria (PKU) can be diagnosed at birth by a blood test. Some conditions that may be present at birth do not generate observable symptoms until the infant is several months to a year or more old. Among these are PKU, Tay-Sachs disease, cystic fibrosis, sickle cell anemia, and hearing loss. Various types of mental retardation, cerebral palsy, and minimal brain dysfunction cannot be observed until the infant begins (or fails to begin) evolving through normal stages of psychomotor development.

Most American hospitals administer a routine test to determine the infant's physical condition at birth. Named after a noted anesthesiologist, the late Virginia Apgar, the APGAR test measures the baby's condition according to five significant variables: "Appearance or coloring," "Pulse," "Grimace or reflex irritability," "Activity," and "Respiration." Any significant variation from the normal response to these measures may require further testing and observation. In later infancy, other indications of normal physical and psychomotor development can be observed; significantly abnormal development may indicate the existence of a birth defect.

Phases of normal infant development*

Laughs—2 months
Rolls over—2.8 months
Reaches out for an object on a table held by mother—3.6 months
Sits alone for at least five seconds—5.5 months.
Says "mama" or "dada"—6.9 months

—Continued

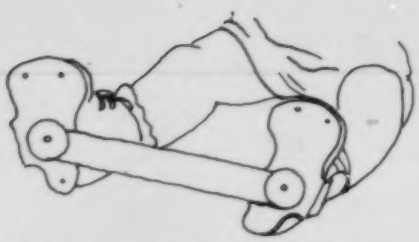
*These examples are selected from the Denver Developmental Screening Scale and represent the normal psychomotor development of 50 percent of the children studied.

Concept 2: There are more than 2,000 types of genetic conditions and birth defects.

Subconcept 2.2—Some defects are manifested at birth; others do not appear until later in life.

2.2.1 (continued)

Many serious congenital diseases and disorders appear during the first two years of life.



Club foot can be corrected by braces.

Picks up a raisin, using thumb and fingers—8.3 months
Plays pat-a-cake—9.1 months
Drinks from a cup—11.7 months
Walks—12.1 months
Copies a drawing of a circle—2.6 years
Recognizes three colors—3 years

Learning Activities

1. Invite a pediatrician or infant care specialist to talk with the class about the physical and mental development of young children.

2. Visit an infant day care center or preschool and find out what measures are used there to determine the children's normal physical and mental development.

Suggested Evaluation Activity

For discussion or writing: How can various genetic conditions that appear during the first two years of life be detected?

2.2.2

Some congenital diseases and disorders occur in later stages of development.

Recommended Grade Level: Intermediate (4–6)

Student Learning Objective

Describe at least two congenital diseases or disorders whose symptoms are not manifested until after the first two years of life.

Content

A baby may appear normal for the first one or two years of life and yet carry the potential for a serious congenital disease or disorder that will not appear until later developmental stages. The effects of Duchenne-type muscular dystrophy are not evident until a child is between two and five years old. Diabetes is rarely manifested before the individual is eight years old. As the body matures into adulthood many disorders and syndromes relating to sexual maturation become evident during adolescence; these include Klinefelter's syndrome and Turner's syndrome (insufficient development of sex organs in males and females, respectively).* Some genetically related disorders usually do not develop until late middle age —for example, gout and Huntington's chorea (slow deterioration of the brain and central nervous system).

—Continued

*Note: Teachers should exercise care in discussing these conditions with preadolescent students; late-maturing students may be unnecessarily alarmed.

Some congenital diseases and disorders occur in later stages of development.

2.2.2
(continued)

Learning Activities

1. Have the students create a chart divided according to varying stages of physical development and age. Working either singly or in groups, the students research particular diseases and disorders and then, as part of a class presentation, place them in the appropriate developmental range on the chart.

2. In discussion, compare the feelings students are willing to express about people who have different types of birth defects. Explore why there are different patterns of acceptance when the defects/disabilities are physical as opposed to mental. Which are easier to accept in one's friends? Relatives? Classmates? Discuss reasons why.

Suggested Evaluation Activity

For writing or class discussion: Indicate which of the following birth defects appear during infancy and which appear in later phases of development:

Duchenne-type muscular dystrophy
Diabetes
Cleft lip/palate
Club foot
Phenylketonuria (PKU)
Cystic fibrosis
Cerebral palsy
Hydrocephalus
Tay-Sachs disease
Huntington's chorea
Klinefelter's syndrome

A child with muscular dystrophy

Notes:

Concept 2: There are more than 2,000 types of genetic conditions and birth defects.

Subconcept 2.3—Birth defects can be the result of a variety of factors.

2.3.1

Factors affecting the mother's health during pregnancy can cause birth defects (poor nutrition, drug or alcohol use, contagious diseases, etc.).

Recommended Grade Level: Intermediate (4–6)

Student Learning Objective

Describe at least two aspects of good prenatal care that affect the health of newborn children.

Key Words

Fetus: The developing organism of a human being prior to birth.

Prenatal care: Maintenance of a mother's health prior to the birth of her child.

Content

Certain factors during pregnancy have been identified as the source of diseases, disorders, and malformations present at birth. Virtually any environmental factor known to have negative effects on a child or adult can have far more serious effects on an unborn infant.

Examples:

Environmental factor	*Congenital disorder*
Rubella (German measles)	Multiple: cataracts, deafness, heart malformation, mental retardation
Radiation	Malformations, genetic defects in future generations
Any drug or medication	Multiple: prematurity, retarded physical and/or mental development
Poor diet	Multiple: prematurity, retarded physical and/or mental development
Undercooked meat, cat feces (toxoplasmosis infection)	Mental retardation, epilepsy, hydrocephalus, eye damage, hearing loss

Learning Activities

1. Discuss: Are there any environmental factors that could harm an unborn baby that would *not* harm any of the students in the class? Examples: poor diet, smoking, radiation, air pollution, alcohol abuse.

2. Have the students describe what they think would be an "ideal" diet for an expectant mother. Compare this with an obstetrician's recommended diet. Resource: "Nutrition and Birth Defects Prevention," Na-

—Continued

Concept 2: There are more than 2,000 types of genetic conditions and birth defects.

Subconcept 2.3—Birth defects can be the result of a variety of factors.

Factors affecting the mother's health during pregnancy can cause birth defects (poor nutrition, drug or alcohol use, contagious diseases, etc.). **2.3.1** (continued)

tional Foundation/March of Dimes pamphlet (see "Agencies" in the reference section).

3. Invite a public health nurse or the school nurse to discuss the effects of rubella during pregnancy and immunization campaigns to prevent the spread of the disease.

Suggested Evaluation Activity

For discussion or writing: What are some things a pregnant woman can do (or avoid doing) to ensure that she will have a healthy baby?

Although premature birth is not itself a defect, it can affect the child's later development. **2.3.2**

Recommended Grade Level: Secondary (7–12) and adult education

Student Learning Objective

Identify at least two causes of premature birth and two possible negative effects of premature birth on the child's later development.

Content

Many different factors can lead to premature delivery: the age of the mother, the number of previous pregnancies, illnesses or poor nutrition during pregnancy, etc. A baby that is undersized or underweight is generally considered to be premature, whether or not it is a full-term child (266 days from conception to birth). Studies have revealed that premature babies are more prone to a variety of problems, including mental retardation and early death, than full-term babies. The most serious concern arising from prematurity is the child's immediate survival. More than half of the infants that die in the first week after birth are premature.

Premature baby in hospital infant intensive care unit

Learning Activities

1. Invite a physician to talk with the class about how a hospital treats premature babies. If possible, arrange for small groups of students to observe a hospital nursery or infant intensive care unit.

2. Use slides or photographs to illustrate the prenatal development of a human fetus and discuss the various stages of development.

Suggested Evaluation Activity

For writing or discussion:

- How is prematurity defined?
- What are some possible consequences of premature birth?

Concept 2: There are more than 2,000 types of genetic conditions and birth defects.

Subconcept 2.3—Birth defects can be the result of a variety of factors.

2.3.3

Birth defects are frequently the result of genetic factors.

Recommended Grade Level: Secondary (7–12) and adult education

Student Learning Objective

Distinguish between genetic factors and other kinds of factors that can cause birth defects.

Key Word

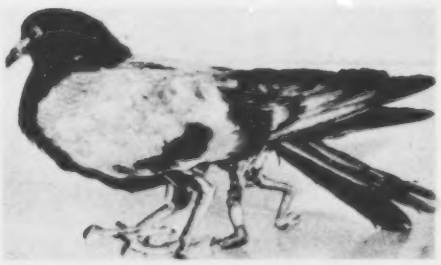
Mutation: A failure of a gene to produce an exact self-copy, resulting in modification of the hereditary trait produced by that gene.

Content

Even after many years of advanced research medical scientists are uncertain of the exact causes of more than half the congenital defects found in humans. Approximately one fifth are known not to be genetically related; these are the defects resulting from such environmental factors as infection, drugs, or physical injury to the fetus. At least one fifth of all other birth defects are known to be the result of genetic factors. The remaining defects may result from either environmental or genetic factors, or a combination of the two. Some genetic diseases and disorders are transmitted genetically from one generation to another; others may be the result of genetic mutations occurring for the first time.

Conditions resulting from:

Environmental factors	*Genetic factors*	*Environmental, genetic, or combination*
Multiple disorders following rubella during pregnancy	Hemophilia	Club foot
Brain damage or mental retardation resulting from drug use	Color blindness	Cleft lip/palate
	Tay-Sachs disease	Premature birth
	Sickle cell anemia	Still birth
	Down's syndrome	



Four-legged bird, the result of a mutation

Learning Activities

1. As the class continues its study of different kinds of genetic diseases and birth defects maintain a chart with three categories of causes: "Environmental factors," "genetic factors," and "environmental, genetic, or combination." Each time the class learns about a new condition or disease discuss where on the chart it belongs and keep a record of what the class decides.

Suggested Evaluation Activity

For discussion or writing: Give an example of a birth defect resulting from genetic factors, a birth defect resulting from environmental factors, and a birth defect resulting from a combination of the two.

Concept 2: There are more than 2,000 types of genetic conditions and birth defects.

Subconcept 2.4—Genetic conditions are transmitted in several different ways.

Some genetic conditions are the result of chromosome abnormalities. 2.4.1

Recommended Grade Level: Secondary (7–12) and adult education

Student Learning Objective

Identify at least two genetic conditions that are the result of chromosome abnormalities.

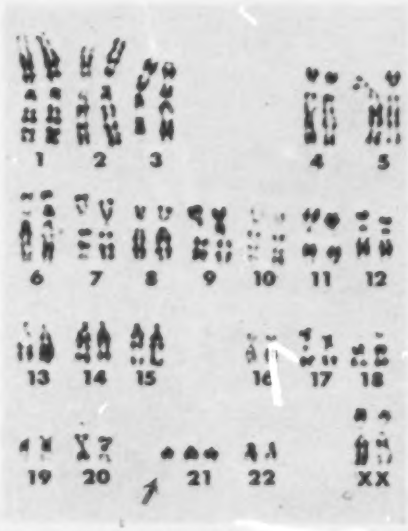
Content

Chromosome abnormalities, or errors, are a principal cause of birth defects. Since the chromosomes, which carry the genes, are the major determinant of inherited characteristics, even the slightest chromosome abnormality involves scores of genes and can cause severe developmental problems in a newly conceived fetus. Most miscarriages in early pregnancy are the result of chromosome errors. Infants with chromosome errors are often severely handicapped.

Any number of factors may cause chromosome errors, but very little is known about the specific causes. Medical geneticists have speculated that such environmental factors as radiation, drugs, and the age of the parents can contribute to chromosome errors.

The most serious chromosome errors are the result of the cells of the sperm, egg, or newly developed fetus containing more or less than the usual amount of chromosomal material. Down's syndrome is the best known result of such an error. Severe birth defects can also occur when parts of chromosomes break off and join on to other chromosomes, thus interfering with the normal transmission of genetic information.

Abnormalities in the sex chromosomes are less severe than those affecting the autosomes. Abnormal numbers of sex chromosomes can produce Klinefelter's syndrome in teenage boys and Turner's syndrome in teenage girls (both conditions involve abnormal sexual maturation). Although less damaging, larger than normal numbers of sex chromosomes can produce unusual height and some mental retardation in both males and females.



Human chromosomes with an additional #21 chromosome, resulting in Down's syndrome

Learning Activities

1. Find pictures of broken or abnormal chromosomes.

2. Discuss the pros and cons of testing female Olympic athletes to determine whether or not they had indications of excessive male sex chromosomes and hormones. The test was required of all participating female athletes in the 1976 Olympics and was made by obtaining a smear from inside the individual's mouth and then examining it under a microscope for the presence of chromatin, a substance found only in female cells.

Suggested Evaluation Activity

For discussion or writing: Down's syndrome is the result of one kind of chromosome abnormality; Klinefelter's syndrome, of another. Describe the two different kinds of chromosome abnormalities that cause these two disorders.

Concept 2: There are more than 2,000 types of genetic conditions and birth defects.

Subconcept 2.4—Genetic conditions are transmitted in several different ways.

2.4.2

Some genetic conditions are the result of one of three single-gene defects: dominant inheritance, recessive inheritance, or X-linked inheritance.

Recommended Grade Level: Secondary (7–12) and adult education

Student Learning Objective

Distinguish between dominant, recessive, and X-linked inheritance, and discuss the chances of a child being affected by an inherited condition in each of these three patterns.

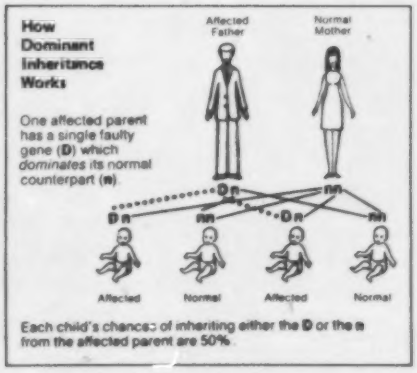
Content

Just as certain harmless physical traits such as hair and eye color may be either dominant or recessive when they are passed to new generations through linked pairs of genes, diseases and disorders may also be either dominant or recessive. Genetic conditions that occur as the result of one harmful gene in a pair are single-gene defects. When they appear on the autosomes, these defects are often called either "autosomal dominant" or "autosomal recessive." A third variety is located on the sex (or X) chromosomes and is therefore known as "sex-linked" or "X-linked."

- *Dominant conditions.* One parent is affected by the condition; the other is not. Each child of these parents has a 50 percent chance of being affected by the condition. These include such conditions as achondroplasia, a form of dwarfism; glaucoma; Huntington's chorea; hypercholesteremia, a cause of heart disease; and polydactyly, extra fingers or toes.

- *Recessive conditions.* These conditions occur when both parents carry the same recessive gene. Although there is only a 25 percent chance that a child from these parents will be affected, there is a 50 percent chance of a child being a carrier. Thus, in later generations the condition may recur if a male carrier and a female carrier produce children. Recessive conditions tend to be more severe than dominant conditions. They include such conditions as cystic fibrosis, galactosemia, phenylketonuria (PKU), sickle cell anemia, and thalassemia.

—Continued



Notes:

Concept 2: There are more than 2,000 types of genetic conditions and birth defects.

Subconcept 2.4—Genetic conditions are transmitted in several different ways.

Some genetic conditions are the result of one of three single-gene defects: dominant inheritance, recessive inheritance, or X-linked inheritance.

2.4.2
(continued)

• *X-linked conditions.* The mother is the carrier of X-linked conditions. While the mother is not affected, she carries one abnormal gene on her sex chromosome and one normal gene. There is a 50 percent risk that each male child will be affected by the disease and a 50 percent risk that each female child will be a carrier. Among the more common X-linked conditions are certain forms of color blindness, hemophilia, and muscular dystrophy.

Learning Activities

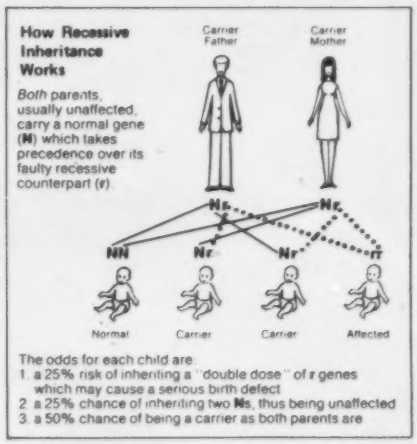
1. For class discussion:
   - Considering what you know about single-gene defects, why do you think that incest and intermarriage among first cousins are considered unhealthy practices?
   - Name several different kinds of racial, ethnic, geographical, or religious groups that tend to intermarry. Are there any diseases associated mainly with these groups? Explain why they are likely to occur.

2. From the school nurse or an ophthalmologist obtain charts to screen for color blindness. Check all students who volunteer. If any students are color blind, ask them to trace this characteristic in their families. Discuss the adaptations color blind people must make to everyday life.

Suggested Evaluation Activity

For discussion or writing: Specify the inheritance patterns for each of the following conditions:

Polydactyly	Cystic fibrosis
Phenylketonuria (PKU)	Huntington's chorea
Hemophilia	Sickle cell anemia
Muscular dystrophy	Color blindness


How Recessive Inheritance Works
Both parents, usually unaffected, carry a normal gene (N) which takes precedence over its faulty recessive counterpart (r).
Carrier Father
Carrier Mother
Nr
Nr
NN
Nr
Nr
rr
Normal
Carrier
Carrier
Affected
The odds for each child are:
1. a 25% risk of inheriting a "double dose" of r genes which may cause a serious birth defect
2. a 25% chance of inheriting two Ns, thus being unaffected
3. a 50% chance of being a carrier as both parents are

Notes:

Concept 2: There are more than 2,000 types of genetic conditions and birth defects.

Subconcept 2.4—Genetic conditions are transmitted in several different ways.

2.4.3

Some genetic conditions are the result of multifactorial inheritance.

Spina bifida, showing spinal cord and protruding sac containing spinal fluid

Recommended Grade Level: Secondary (7–12) and adult education

Student Learning Objective

Define multifactorial inheritance, and identify at least two conditions associated with it.

Content

Multifactorial inheritance does not follow any of the mathematically precise Mendelian patterns of single-gene inheritance. It is distinct from chromosome abnormalities, however, in that it is hereditary, involving the interaction of genes with one another or with environmental factors. The incidence of multifactorial conditions varies; some conditions are considered multifactorial in one population but not in another. For example, a condition known as spina bifida, a defect in the spinal cord, occurs in about 1 in 1,000 live births in the United States, where it is probably not multifactorially inherited. In England, among the Welsh, it occurs in 1 out of 600 live births, and an actual inheritance pattern is more likely. Some defects generally considered to be the result of multifactorial inheritance are cleft lip/palate, club foot, congenital dislocation of the hip, and hydrocephalus.

Learning Activities

1. Ask for volunteers to research, and report to the class about, conditions that are multifactorial in one population but not in another.

2. Refer to a dictionary, encyclopedia, or resource person to define and describe such conditions as spina bifida and hydrocephalus.

Suggested Evaluation Activity

For discussion or writing: When do you think spina bifida or cleft lip/palate would or would not be the result of multifactorial inheritance?

Notes:

3

Goal 3: To acquire basic information about the prevention and treatment of genetic conditions and birth defects and to know where to seek help and further information.

Concept 3: Some genetic conditions and birth defects can be prevented, treated, or alleviated.

Goal 3: To acquire basic information about the prevention and treatment of genetic conditions and birth defects and to know where to seek help and further information.

Concept 3: Some genetic conditions and birth defects can be prevented, treated, or alleviated.

Subconcept	Recommended grade level for instruction*		
	Primary (K–3)	Intermediate (4–6)	Secondary (7–12) & adult education
3.1—Treatment is available both for relatively minor genetic conditions and for conditions that have serious, long-term effects.			
3.1.1—Minor conditions such as poor vision can be corrected or alleviated.	●	○	○
3.1.2—Diseases and disorders such as Rh incompatibility and diabetes can be prevented or treated.		●	○
3.1.3—Certain genetic conditions require extensive medical, educational, or therapeutic treatment.		●	○
3.2—There are several ways of diagnosing or predicting genetic conditions and birth defects.			
3.2.1—Individuals should be acquainted with their family's medical history.	●	○	○
3.2.2—Amniocentesis and other tests have enabled physicians to detect the presence of a number of birth defects prior to birth or immediately after birth.		●	○
3.2.3—Programs dealing with genetic conditions and birth defects, particularly genetic counseling programs, have expanded dramatically in recent years.			●
3.2.4—Many different options are available to prospective parents who believe that their child may have a genetic condition or birth defect.			●

*Note: A solid bullet (●) indicates that this grade level is recommended for teaching this particular subconcept. An open bullet (○) indicates that the subconcept should have been mastered by students at this grade level. Since many students have not received instruction about genetic conditions and birth defects in the earlier grades, instruction may be required at more advanced grade levels for concepts recommended for earlier levels. These recommendations are not intended to be followed rigorously; rather, they are an outline of how a course of study in genetic conditions and birth defects might progress from one grade level to another.

Concept 3: Some genetic conditions and birth defects can be prevented, treated, or alleviated.

Subconcept 3.1—Treatment is available both for relatively minor genetic conditions and for conditions that have serious, long-term effects.

Minor conditions such as poor vision can be corrected or alleviated. 3.1.1

Recommended Grade Level: Primary (K–3)

Student Learning Objective

Describe at least two treatable minor genetic conditions or birth defects and the means of treatment for each.

Content

Strabismus ("crossed eyes") affects two percent of the population with varying degrees of severity. Three percent of all infants have birthmarks. Recognizing that conditions such as these are relatively common can prepare students to accept and understand birth defects that have more damaging consequences.

Examples:

Condition	*Treatment*
Strabismus (crossed eyes)	Glasses, eye patch, eye exercises, surgery
Amblyopia (weak eye)	Glasses, eye patch, eye exercises
Birthmarks	Usually none required, or cosmetic surgery
Moles	Removal under local anesthetic

Learning Activities

1. Conduct a class survey and list the following information on a chart:

- How many students have:
  - Poor vision
  - Birthmarks
  - Any other minor condition that was present at birth
- How many students have ever had a similar congenital condition that was treated?
- How many students have a close relative with such a condition?
- How many students have ever known anyone with such a condition?

2. Develop a "Treatable Birth Defects" poster, depicting the conditions and treatments with photographs or drawings.

Suggested Evaluation Activity

For discussion or writing: What kinds of treatment are available for:

Strabismus
Amblyopia
Birthmarks
Moles

Concept 3: Some genetic conditions and birth defects can be prevented, treated, or alleviated.

Subconcept 3.1—Treatment is available both for relatively minor genetic conditions and for conditions that have serious, long-term effects.

3.1.2

Diseases and disorders such as Rh incompatibility and diabetes can be prevented or treated.

Recommended Grade Level: Intermediate (4–6)

Student Learning Objective

Identify at least three treatable congenital diseases or disorders and describe the methods of treating them.

Content

Some congenital diseases and disorders are fatal—Tay-Sachs disease is a notable example—and no way of treating them has yet been found. The recent expansion of research into genetic disease and birth defects has produced ways of treating and preventing numerous other congenital defects that might once have had extremely serious consequences.

Examples:

Condition	*Treatment*	*Prevention*
Prematurity	Intensive care, high nutrition diet	
Cleft lip/palate	Corrective surgery, speech therapy	
Diabetes	Special diet, insulin	
Rh incompatibility	Blood transfusions	Rh vaccine
Club foot	Corrective surgery, corrective braces or casts	
Congenital heart disease	Corrective surgery	
Hemophilia	Blood plasma concentrates	
Retarded physical and mental development resulting from rubella		Vaccination of the mother-to-be
Phenylketonuria (PKU)	Special diet	

—Continued

After surgery to correct cleft lip

Notes:

Concept 3: Some genetic conditions and birth defects can be prevented, treated, or alleviated.

Subconcept 3.1—Treatment is available both for relatively minor genetic conditions and for conditions that have serious, long-term effects.

Diseases and disorders such as Rh incompatibility and diabetes can be prevented or treated. **3.1.2** (continued)

Learning Activities

1. Invite an adult with either diabetes or hemophilia to discuss his or her condition with the class.
2. Research project. Using an encyclopedia as a resource, compare the ways in which hemophilia affected the lives of the British royal family in the nineteenth century with the way a modern family might cope with the disease.
3. Discuss: After comparing the diseases in this section, all of which are serious but treatable, and assuming you had a "forced choice," which one would you select? What reasons can you give for your choice?

Suggested Evaluation Activity

For discussion or writing: What congenital disease or disorder might a person have who requires any one of the following kinds of treatment?

- Insulin injections
- Blood plasma concentrates
- Any kind of special diet

Certain genetic conditions require extensive medical, educational, or therapeutic treatment. **3.1.3**

Recommended Grade Level: Intermediate (4–6)

Student Learning Objective

Describe at least two long-term genetic conditions or birth defects, and suggest alternative ways of treating them.

Content

There are an estimated three million mentally retarded individuals in America today. Half a million people now living were born completely or partially blind. Hearing problems and muscular disorders are also birth defects that hundreds of thousands of people must learn to live with. These are all conditions that may affect their victims throughout their lives.

While there has been a great deal of attention to the problem of children with major learning disabilities and developmental problems, educators and pediatricians are far from unanimous about the methods of treatment. Some educators advocate the "mainstreaming" of such children, helping them to adapt to a normal school environment by joining classes of normal children as much as possible. At the other extreme, special classes for the "educable mentally retarded" and, on occasion, institutionalization, have been a recommended approach. There is extensive research evidence that mentally retarded children develop more fully in a family setting than in institutions.

—Continued

Concept 3: Some genetic conditions and birth defects can be prevented, treated, or alleviated.

Subconcept 3.1—Treatment is available both for relatively minor genetic conditions and for conditions that have serious, long-term effects.

3.1.3 (continued)

Certain genetic conditions require extensive medical, educational, or therapeutic treatment.

Learning Activities

1. Invite a specialist in learning disabilities to talk to the class about how various disabilities are diagnosed and treated. Resource: California State Department of Health regional centers for the prevention and treatment of developmental disabilities (see "Agencies" in the reference section).

2. Have one group of students pretend they are deaf or blind. Have another group of students attempt to teach something to the first group. Use blindfolds to simulate blindness and ear plugs or earphones with static noise to simulate deafness.

3. For advanced intermediate or secondary students: Have a classroom debate; each side represents one of the following positions:

- Mentally retarded children should be separated as much as possible from other children, and those with IQs below a certain level should be institutionalized.
- As much as possible, mentally retarded children should have the same kinds of experience, both at home and at school, as other children.

Suggested Evaluation Activity

For discussion or writing: What alternatives are there for treating a mentally retarded child?

Notes:

Concept 3: Some genetic conditions and birth defects can be prevented, treated, or alleviated.

Subconcept 3.2—There are several ways of diagnosing or predicting genetic conditions and birth defects.

Individuals should be acquainted with their family's medical history. 3.2.1

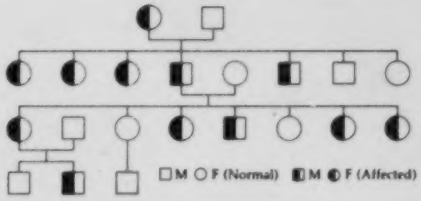
Recommended Grade Level: Primary (K–3)

Student Learning Objective

State at least two reasons why it is important to know about the medical history of one's family.

Content

Numerous diseases and disorders that may occur during an individual's development can be anticipated and treated more effectively, or even prevented, when the individual recognizes that such disorders have occurred before in his or her family. Considering the importance of genetic factors to personal health, ignorance about one's own personal and family medical history can create problems not only for oneself but also for one's children. For adults who wish to become parents a family medical record is an important component of family planning. While children would not ordinarily keep such a record, an awareness of their own and their family's medical history can prepare them for the larger responsibilities of adulthood.



A family medical pedigree, showing transmission of a genetic condition

Learning Activities

1. Have the students fill out the following personal medical record checklist:

Either I or someone in my immediate family has had one or more of the following illnesses or disorders:

	I	Others in my family
Allergies	_____	_____
Arthritis	_____	_____
Cancer	_____	_____
Diabetes	_____	_____
Epilepsy	_____	_____
Hearing defects	_____	_____
Heart disease	_____	_____
Mental retardation	_____	_____
Obesity	_____	_____
Tuberculosis	_____	_____
Visual defects	_____	_____
Other _____	_____	_____

—Continued

Concept 3: Some genetic conditions and birth defects can be prevented, treated, or alleviated.

Subconcept 3.2—There are several ways of diagnosing or predicting genetic conditions and birth defects.

3.2.1
(continued)

Individuals should be acquainted with their family's medical history.

Discuss:

- Which of these diseases or disorders are contagious?
- Which might be considered congenital?

2. Discuss the following questions:
   - Are there any health problems in my family that might affect me in the future?
   - Could my health and my family's be improved by better health practices?

Suggested Evaluation Activity

For discussion or writing: Why is it important for you to know about your family's medical history.

Notes:

Concept 3: Some genetic conditions and birth defects can be prevented, treated, or alleviated.

Subconcept 3.2—There are several ways of diagnosing or predicting genetic conditions and birth defects.

Amniocentesis and other tests have enabled physicians to detect a number of birth defects prior to birth or immediately after birth. 3.2.2

Recommended Grade Level: Intermediate (4–6)

Student Learning Objective

Describe at least two methods of testing for congenital defects prior to, or immediately after, birth and at least one disease that can be diagnosed through each method.

Content

Amniocentesis is a medical procedure that enables a physician to examine the chromosomes and to study the body chemistry of an unborn child while it is still in the mother's womb. Usually performed after the fourteenth week of pregnancy, amniocentesis is accomplished by inserting a syringe needle through the abdominal wall into the amniotic fluid that surrounds the developing fetus. Cells from the fluid are then grown in cultures that can be analyzed for their chromosomal content. Among several serious genetic disorders that can be detected through amniocentesis are Tay-Sachs disease and Down's syndrome. One incidental benefit of amniocentesis is that it can be used to identify the sex of the unborn fetus; this can be helpful to families with sex-linked (male only) disorders.

Other tests often administered prior to, or immediately after, birth:

- Blood tests to identify blood diseases such as sickle cell anemia and thalassemia.
- Enzyme tests of fetal blood cells to identify such metabolic disorders as diabetes or phenylketonuria (PKU).

Learning Activities

1. Examine drawings or photographs of amniocentesis.
2. Inquire at a local hospital how many women ordinarily have amniocentesis in a given month; compare this with the same month five or ten years ago.
3. Conduct a survey of all the pregnant women whom students in the class know to find out how many of them know about amniocentesis.

Suggested Evaluation Activity

For discussion or writing: Which of the following can be detected through amniocentesis?

Thalassemia	Phenylketonuria (PKU)
Sickle cell anemia	Tay-Sachs disease
Down's syndrome	Hemophilia

Amniocentesis

Concept 3: Some genetic conditions and birth defects can be prevented, treated, or alleviated.

Subconcept 3.2—There are several ways of diagnosing or predicting genetic conditions and birth defects.

3.2.3

Programs dealing with genetic conditions and birth defects, particularly genetic counseling programs, have expanded dramatically in recent years.

Recommended Grade Level: Secondary (7–12) and adult education

Student Learning Objective

Identify at least two aspects of genetic counseling and at least one source of genetic counseling.

Content

Genetic counseling

With the development of genetics as a science between 1900 and 1950 came new knowledge about genetically related diseases and disorders and new research techniques for diagnosing, preventing, and treating such conditions. This knowledge has produced a wide expansion of programs to deal with genetic conditions and birth defects.

More than a dozen states have implemented massive screening programs for such diseases as sickle cell anemia. Forty-eight states screen all newborn infants for phenylketonuria (PKU). Both kinds of screening can be done with a simple blood test.

The number of genetic counseling programs throughout the country has grown from 13 in 1955 to several hundred today. Usually located in hospitals and medical centers, genetic counseling programs offer prospective parents an opportunity to discuss their family's medical history and to undertake tests for various genetic diseases administered by qualified physicians and geneticists. Genetic counseling is commonly sought by parents who have already had at least one child with a genetic disease or disorder.

Genetic counseling services are usually provided by professionals who are knowledgeable about both genetics and pediatric medicine. Since a genetic counselor can predict only a limited number of actual birth defects with certainty, for the most part the counselor can only advise the prospective parents about the chances of their children suffering from defects. The genetic counselor's role is to inform and advise, not to make decisions about which course of action the prospective parents should take.

Learning Activities

1. Invite a genetic counselor from a local hospital or medical center to talk with the class about the functions of genetic counseling.
2. Discuss: Who should have genetic counseling?
3. Invite a speaker to talk with the class about the prevention of birth defects. Resource: Local National Foundation/March of Dimes chapter (see "Agencies" in the reference section).

Suggested Evaluation Activity

For discussion or writing: Describe two kinds of assistance that prospective parents can expect from genetic counseling.

Concept 3: Some genetic conditions and birth defects can be prevented, treated, or alleviated.

Subconcept 3.2—There are several ways of diagnosing or predicting genetic conditions and birth defects.

Many different options are available to prospective parents who believe that their child may have a genetic condition or birth defect. 3.2.4

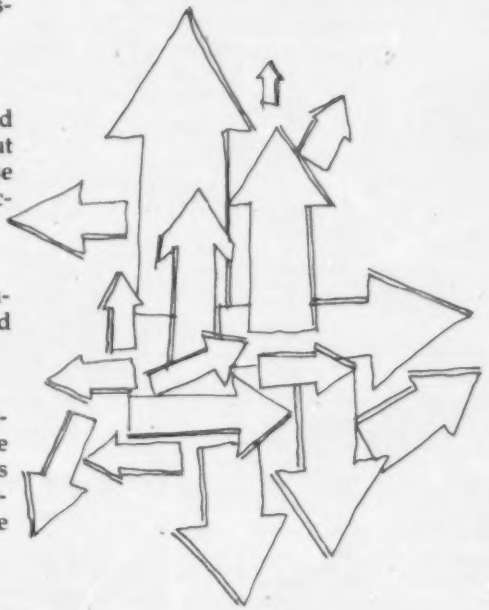
Recommended Grade Level: Secondary (7–12) and adult education

Student Learning Objective

Describe at least two options available to prospective parents who discover that their child may have a genetic condition or birth defect.

Content

Prospective parents who are knowledgeable about genetic conditions and birth defects may wish to seek help or advice if they are concerned about having children with congenital problems. In most cases there is no cause for concern. In cases where congenital problems may exist, the prospective parents may choose one of several options:



- *Terminate pregnancy.* In the event of a certain diagnosis of Down's syndrome, Tay-Sachs disease, or other conditions, some parents would find this course a compelling one.

- *Risk having a child with a birth defect.* Amniocentesis can accurately predict many severe birth defects. A genetic analysis of the prospective parents' family medical histories, however, can predict only the chances of a genetic disease—a 25 percent chance of a recessive defect or a 50 percent chance of a dominant condition. Many parents would rather take the risk than terminate a pregnancy.

- *Artificial insemination.* If the parents are advised in genetic counseling that they face significant risks of transmitting a genetic defect to their children, it is possible that the mother can still bear a child after the semen from a healthy father is artificially planted in her uterus. The success rate of artificial insemination is about 20 percent.

- *Adoption.* Some parents may choose not to conceive their own biological children if they consider the risks of birth defects too severe. Such parents could still have a family by adopting children. Declining birth rates and an increasing number of single mothers raising their own families, however, have made it difficult for prospective adoptive parents to find healthy infants for adoption.

- *Have no children.* Seventy-five years ago not having children would have been inconceivable to most married couples. Today, with overpopulation a major social concern and the growing tendency of both parents to pursue careers outside the home, childless couples are more common.

—Continued

Concept 3: Some genetic conditions and birth defects can be prevented, treated, or alleviated.

Subconcept 3.2—There are several ways of diagnosing or predicting genetic conditions and birth defects.

3.2.4 (continued)

Many different options are available to prospective parents who believe that their child may have a genetic condition or birth defect.

Learning Activities

1. Set up a role-play situation in which two people who are prospective parents go to see a genetic counselor. Have two groups portray the following situations. After the role play allow time for class discussion.

   - The counselor explains that the results of amniocentesis indicate that the child will have Down's syndrome; the counselor asks the parents to consider what they will do.
   - The counselor tells the parents that their unborn child will have a 25 percent chance of being affected by cystic fibrosis. The parents discuss what to do.

Suggested Evaluation Activity

For discussion or writing: What would you do if you were a parent and learned through genetic counseling that your unborn child had:

- An almost certain chance of having Down's syndrome
- A 25 percent chance of having cystic fibrosis
- A 50 percent chance of having hemophilia

Notes:

4

Goal 4: To understand the personal, social, and economic consequences of genetic conditions and birth defects and to gain insight into the important ethical questions raised by advances in genetic medicine.

Concept 4: Genetic conditions and birth defects affect individuals, families, and the society.

Goal 4: To understand the personal, social, and economic consequences of genetic conditions and birth defects and to gain insight into the important ethical questions raised by advances in genetic medicine.

Concept 4: Genetic conditions and birth defects affect individuals, families, and the society.

Subconcept	Recommended grade level for instruction*		
	Primary (K–3)	Intermediate (4–6)	Secondary (7–12) & adult education
4.1—It is important to be able to accept genetic conditions in oneself and in others.			
4.1.1—All people are born with some features or qualities they like.	●	○	○
4.1.2—All people are born with some features or qualities they would prefer not to have.	●	○	○
4.2—Genetic conditions and birth defects are extremely costly to society.			
4.2.1—Some of the costs are emotional in nature, principally to the affected individual and his or her family		●	○
4.2.2—Some of the costs are financial.		●	○
4.3—Throughout history there have been attempts to develop "ideal" human beings.			
4.3.1—Philosophers, scientists, and political leaders have periodically advocated the use of eugenics as a way of improving human beings.			●
4.3.2—Important recent advances have been made in the technology of genetic engineering (genetic counseling, "test tube" babies, cloning).			●
4.3.3—Our society is on the threshold of being able to determine the kinds of people that are born, and this has profound moral and ethical implications.			●

*Note: A solid bullet (●) indicates that this grade level is recommended for teaching this particular subconcept. An open bullet (○) indicates that the subconcept should have been mastered by students at this grade level. Since many students have not received instruction about genetic conditions and birth defects in the earlier grades, instruction may be required at more advanced grade levels for concepts recommended for earlier levels. These recommendations are not intended to be followed rigorously; rather, they are an outline of how a course of study in genetic conditions and birth defects might progress from one grade level to another.

Concept 4: Genetic conditions and birth defects affect individuals, families, and the society.

Subconcept 4.1—It is important to be able to accept genetic conditions in oneself and in others.

All people are born with some features or qualities they like. 4.1.1

Recommended Grade Level: Primary (K–3)

Student Learning Objective

Identify at least two inherited traits or characteristics in yourself that you like.

Content

Self-acceptance is a prerequisite for accepting and understanding others, particularly others who may be perceived as physically or mentally inferior. This concept is deceptively simple; the learning activities may require considerable sensitivity and tactful assistance on the part of the teacher if some students appear to have negative self-concepts.

Learning Activities

1. Have each student stand for a short time in front of a full-length mirror. Then ask the students to write a brief statement about "Things I Saw That I Liked."

2. Divide the class into small groups. Each group discusses the topic "My Best Qualities." After the students have identified several qualities in themselves that they consider their "best," ask them to rank the qualities on a spectrum ranging from "probably, or definitely, inherited from my parents" to "probably, or definitely, the result of my environment."

3. Ask for volunteers to tell the class their reactions to the following statements:

- The quality of my mother that I like best and also have is . . . .
- The quality of my father that I like best and also have is . . . .

Suggested Evaluation Activity

Discuss: Why do you think people who like themselves might be happier than people who dislike themselves? What makes a person like himself or herself?

Notes:

Concept 4: Genetic conditions and birth defects affect individuals, families, and the society.

Subconcept 4.1—It is important to be able to accept genetic conditions in oneself and in others.

4.1.2

All people are born with some features or qualities they would prefer not to have.

Recommended Grade Level: Primary (K–3)

Student Learning Objective

Describe several ways of enabling oneself and others to accept a defect or disorder.

Content

The way we react to physical defects or disorders in others is often determined by the way we feel about ourselves. Similarly, our own imperfections can be the source either of anguish or of strength. This concept can lead in many different directions. The focus may be either on physical qualities or qualities of character, or both. For teachers who are comfortable dealing with group dynamics physical defects can be a touchstone for investigating the motives behind intergroup rivalries and cruel behavior among students.

Learning Activities

1. Have each student privately note down a response to "The one quality in myself I would change if I could is . . . .". The students may share their responses with the rest of the class if they wish. Ask the students to write a private response to the following question: "I probably can't change this one quality. Therefore, ..." Again, students may share their responses on a voluntary basis.

2. Read a biography of Helen Keller.

3. Have a class discussion about teasing. Introduce the following questions:

   - Who engages in teasing?
   - Why do people tease other people?
   - What kinds of things do people tease others about?
   - Instead of teasing someone with a particular weakness what would be a way of making that person feel more accepted?

4. Have the students discuss or write about their feelings in response to the following situation: You pass a crippled person on the street. What is your reaction?

Suggested Evaluation Activity

Discuss: What things could you do to help a disabled schoolmate feel more comfortable or accepted?

Concept 4: Genetic conditions and birth defects affect individuals, families, and the society.

Subconcept 4.2—Genetic conditions and birth defects are extremely costly to society.

Some of the costs are emotional in nature, principally to the affected individual and his or her family.

4.2.1

Recommended Grade Level: Intermediate (4–6)

Student Learning Objective

Analyze the emotional adjustment required of the individual and the family to a serious congenital defect.

Key Words

Sibling: A brother or sister.

Martyrdom: Great suffering, often deliberately chosen.

Content

In the case of birth defects that produce a long-term condition requiring extended treatment and care, an important emotional adjustment is required on the part of both the individual and the family. Common day-to-day emotional consequences of such birth defects include:

- *Guilt.* Both the child and the parents tend to feel that there is something "wrong" or "inferior" about themselves.
- *Denial.* For years the parents may deny, either consciously or subconsciously, that a child is retarded, for example.
- *Over-protectiveness and over-compensation.* Parents of hemophiliacs, for example, frequently try to protect their children from injury, thus stultifying the child's normal psychological need for independence.
- *Problems with siblings.* Often a child with a birth defect is given more attention than his or her siblings, thus producing a higher than normal amount of sibling rivalry. Siblings themselves may experience severe guilt feelings because they are so much "better" than their afflicted brother or sister.
- *Anger.* The parents, the child, and siblings normally have feelings of anger and resentment about their situation. Yet this anger is difficult to express; it can become a permanent influence on the emotional climate of the family. Occasionally this anger can lead to physical abuse of the affected child.
- *Martyrdom.* Parents and affected individuals feel that they must devote all their attention to the problems caused by a particular birth defect even if this is not completely necessary. Their behavior is an indirect expression of their anger and resentment, and it can have only negative effects.

Emotionally healthy acceptance of a genetic condition or birth defect can be difficult to achieve. Often individual or family counseling may be required.

—Continued

Concept 4: Genetic conditions and birth defects affect individuals, families, and the society.

Subconcept 4.2—Genetic conditions and birth defects are extremely costly to society.

4.2.1 (continued)

Some of the costs are emotional in nature, principally to the affected individual and his or her family.

Learning Activities

1. Divide the class into groups. Assign each group a particular role playing situation involving the presence of a disabled child in a family. Allow time for class discussion.

Examples:

- The father resents the child but tries to hide it, thus refusing to deal with his resentment.
- The mother and father over–protect the child.
- The brothers and sisters begin to feel that they no longer receive sufficient attention from their parents.
- The brothers and sisters explain to friends that they have a brother or sister with a birth defect.

2. "Forced choice" exercise. Ask the students to answer the following questions in writing (not to be handed in):

Which would you rather have:

A parent who is too strict	One who pays no attention to you
A brother who is mentally retarded	A brother who is blind
A father with a heart condition	A father with diabetes

Go through some sample answers, and ask different students to state the reasons for their answers.

A mother and her four-week-old child with Down's syndrome

Suggested Evaluation Activity

Discuss: What do you think would be the most difficult problems for the parents of a severely disabled child? For the brothers and/or sisters?

Notes:

Concept 4: Genetic conditions and birth defects affect individuals, families, and the society.

Subconcept 4.2—Genetic conditions and birth defects are extremely costly to society.

Some of the costs are financial. 4.2.2

Recommended Grade Level: Intermediate (4–6)

Student Learning Objective

Analyze the financial cost of a genetic condition or birth defect to the individual or family.

Content

The financial burden of some congenital conditions and disorders can strain family resources far beyond normal limits. A child with Tay-Sachs disease, for example, will inevitably die at the end of two or three years; yet intensive care during this time can cost as much as $35,000. Maintaining a severely retarded individual in an institution can cost between $250,000 and $750,000 over the individual's lifetime. Treatment (mainly dietary) for a child with early stages of phenylketonuria (PKU) can cost more than $10,000; yet treatment can prevent the far more costly effects of severe mental retardation that could occur without it. Since all of the more serious genetic conditions and birth defects require long-term medical care, all of them become financially costly to individuals, families, and the society.

Learning Activities

1. Have the students do a consumer-oriented study of the costs of medical care in your community for a wide variety of genetic conditions and birth defects, ranging from relatively mild defects (poor vision) to the most serious (mental retardation, requiring institutionalization).

Compare the expenses:

- To a family
- To health agencies
- To taxpayers

Suggested Evaluation Activity

For discussion or writing: Which of the following conditions in a child would probably place the greatest financial burden on a family?

- Early stages of phenylketonuria (PKU)
- Down's syndrome
- Advanced stages of PKU
- Hemophilia

Concept 4: Genetic conditions and birth defects affect individuals, families, and the society.

Subconcept 4.3—Throughout history there have been attempts to develop "ideal" human beings.

4.3.1

Philosophers, scientists, and political leaders have periodically advocated the use of eugenics as a way of improving human beings.

Speech therapy following surgery for cleft lip

Recommended Grade Level: Secondary (7–12) and adult education

Student Learning Objective

Describe eugenics, and cite at least two instances in which political leaders attempted to implement eugenics as a social policy.

Key Words

Aryan: Nordic, blond, fair-skinned.

Eugenics: Improving human beings by the deliberate selection of certain types of people to perpetuate their qualities.

Euthenics: Improving human beings through changes in the environment.

Content

The nineteenth-century biologist Francis Galton coined the term "eugenics" to describe the deliberate selection of certain types of human beings to perpetuate their qualities in future generations—in short, breeding better people. "Euthenics"—improving human beings through improvements in the environment—is the opposite.

While not labeled as such, eugenics was envisioned centuries before Galton by the Greek philosopher Plato. The notion of a superior kind of human being, the "philosopher king," was central to Plato's vision of an ideal society in the *Republic*. Plato wrote: "It follows . . . that the best of both sexes ought to be brought together as often as possible, and the worst as seldom as possible and that the issue of the former unions ought to be reared, and that of the latter abandoned if the flock is to attain to first-rate excellence."

Friederich Nietzsche, a nineteenth-century German philosopher and poet, amplified this idea by suggesting that ". . . man is a rope stretched

—Continued

Notes:

Concept 4: Genetic conditions and birth defects affect individuals, families, and the society.

Subconcept 4.3—Throughout history there have been attempts to create "ideal" human beings.

Philosophers, scientists, and political leaders have periodically advocated the use of eugenics as a way of improving human beings.

4.3.1
(continued)

between the animal and the Superman." Nietzsche envisioned the Superman as a perfect being of superior mental and physical ability.

Any actual implementation of the concept of eugenics implies total control of human reproduction by the government or state. In ancient Sparta this was a practical reality; the government determined that only the strongest and most able young men could father children. Closer to memory is Adolf Hitler's attempt to eradicate the Jews during World War II as part of a massive crusade to breed a superior "Aryan" race. More than six million Jews were exterminated in German concentration camps between 1939 and 1945.

Note: This subconcept is not intended as an endorsement of eugenics. The teacher should also present the idea that individual, ethnic, and racial differences are not only desirable, but necessary, for the health and well being of a society.

Adolf Hitler

Learning Activities

1. Have a class debate for and against eugenics.
2. Discuss: Why are individual differences among people important to a society? Do you agree that individual differences *are* important?
3. Ask for volunteers to read, and report to the class about, *The Diary of Anne Frank*, the story of a German Jewish teenager during World War II, or Aldous Huxley's *Brave New World*, the story of a society in which eugenics is the official government policy.
4. Arrange for the students to view either *Judgment at Nuremburg* or *QB VII*. Both films deal with the plight of the Jews during World War II. (Check local theatres, television listings, or film rental services.)

Suggested Evaluation Activity

For discussion or writing: Distinguish between eugenics and euthenics.

Notes:

Concept 4: Genetic conditions and birth defects affect individuals, families, and the society.

Subconcept 4.3—Throughout history there have been attempts to develop "ideal" human beings.

4.3.2

Important recent advances have been made in the technology of genetic engineering (genetic counseling, "test tube" babies, cloning).

Recommended Grade Level: Secondary (7–12) and adult education

Student Learning Objective

Identify at least two technological aspects of genetic engineering with which scientists are currently experimenting.

Content

On the one hand, genetic engineering—the technological manipulation of heredity—has the ring of science fiction, seemingly improbable and unattainable. On the other hand, genetic engineering is an ancient practice.

For centuries farmers and horsebreeders, among others, have experimented with creating new kinds of plants and animals by inbreeding and cross-breeding. Couples who choose not to have children for fear of passing on diseases that are known to "run" in their families often do so without the benefit of genetic counseling or sophisticated laboratory analysis of their chromosomes and blood cells. Both are relatively primitive examples of genetic engineering.

The "new genetics" is distinguished by the extent to which modern scientific experiments have progressed toward determining genetic factors artificially in a laboratory setting.

The first successful experiments with artificial insemination of humans occurred in 1884. "Test tube babies"—the creation of fetuses in artificial wombs—have been the subject of extensive laboratory experimentation for more than a decade. Although no fetus has ever been born from this process, Russian scientists have been able to keep a fetus alive in an artificial womb for more than six months.

Cloning, the reproduction of living organisms from single cells of those organisms, producing a duplicate of the original organism asexually,

—Continued

Notes:

Important recent discoveries have been made in the technology of genetic engineering (genetic counseling, "test tube" babies, cloning).

4.3.2
(continued)

has already been accomplished with plants and some lower orders of animals. One scientist has predicted that a human clone will be produced by 1980; others claim that human clones may not be feasible for decades, or even a century. Popularly this process has been labeled, in speculative discussions, "xeroxing human beings."

Perhaps most important, in the years since the discovery of the structure of the DNA molecule, scientists have conducted laboratory experiments indicating that DNA molecules from different species can be recombined, producing entirely new forms of life. For two years (1974–76) the National Institutes of Health imposed a moratorium on all research in this field, fearing that new virus-like microorganisms might be developed and accidentally released into the population. Strict guidelines for such research were issued in the spring of 1976, and it is now continuing. In the future scientists may be able to produce new genetic codes in a laboratory setting.

Genetic counseling and laboratory experiments have made the eventual control of human heredity a realistic outcome. The main question is not whether such a development is possible—clearly it has begun to occur—but when it will happen.

Learning Activities

1. For class discussion:
   - As a class, decide which current hero would be the most desirable to "carbon copy" (clone), and why. Why might cloning not be a desirable scientific development?

Suggested Evaluation Activity

For discussion or writing: Distinguish between a simple, primitive form of genetic engineering and a more advanced, technological form.

Notes:

Concept 4: Genetic conditions and birth defects affect individuals, families, and the society.

Subconcept 4.3—Throughout history there have been attempts to develop "ideal" human beings.

4.3.3

Our society is on the threshold of being able to determine the kinds of people that are born, and this has profound moral and ethical implications.

Recommended Grade Level: Secondary (7–12) and adult education

Student Learning Objective

Analyze moral and ethical issues resulting from the new genetics.

Content

Although our society is only on the threshold of a genetic revolution, recent scientific discoveries suggest that when the technology of controlling genetics is finally perfected, the human race will rapidly move into one of the most significant stages of history. The moral and ethical questions raised by such a prospect are far-reaching. The dilemma has been aptly described by Theodosius Dobzhansky, a noted geneticist and writer: " . . . if we enable the weak and the deformed to live and to propagate their kind, we face the prospects of a genetic twilight; but if we let them die or suffer when we can save or help them, we face the certainty of a moral twilight."

The implications of the new genetics can best be represented in the form of several key questions:

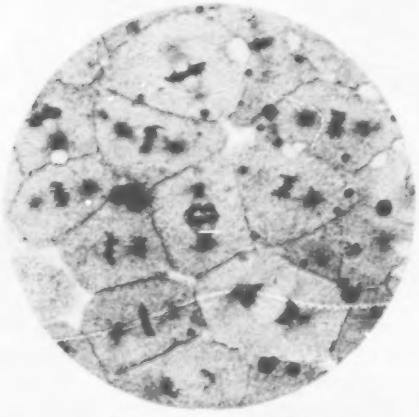
- Who would determine what kinds of people would be born?
- In the event of total control of human reproduction by the state what rights would individuals, both living and unborn, have?
- Is scientific progress inevitable? Should some kinds of scientific experimentation be stopped?

Learning Activities

1. Select volunteers to report to the class about the role of genetics in different science fiction films or books. In these visions of the future what kinds of people exist, and how did they come into being?
2. Have a class debate on the following topic:
   - It is 1985. The government of the state of California has just proclaimed that no one with a genetic disease will be allowed to have children.
   - Argue for and against this policy.

Suggested Evaluation Activity

For discussion or writing: Describe two moral or ethical issues implied by the "new genetics."



Cell division, whitefish

Reference

Glossary

Amino acids. The building blocks of protein, for which DNA forms the genetic code.

Amniocentesis. Needle puncture of the uterus and amniotic cavity through the abdominal wall to allow amniotic fluid to be withdrawn by syringe. The term is often applied to the whole procedure of prenatal diagnosis by culture and analysis of amniotic fluid cells.

Autosome. Any chromosome other than the sex chromosomes. Humans have 22 pairs of autosomes.

Aryan. Nordic, blond, fair-skinned.

Birth defect. A disease, disorder, or other condition present at birth that can impair an individual's health.

Cell. The living active microscopic unit of all plants and animals, consisting of many specialized parts.

Chromosome abnormality, or error. An abnormality of chromosome number or structure.

Chromosomes. Microscopic threadlike bodies in the nuclei of cells.

Clone. A cell line derived by mitosis from a single ancestral cell.

Congenital trait. Trait present at birth, not necessarily genetic.

Cytology. The study of cells.

Cytogenetics. The study of the genetic effects of cellular reproduction, focusing primarily on the chromosomes.

DNA. Deoxyribonucleic acid, the nucleic acid of the chromosomes.

Dominance. The quality of a particular trait that appears when it is paired genetically with a different trait, the second trait being recessive.

Environment. The conditions or influences present in the world an individual inhabits.

Eugenics. Improving human beings by the deliberate selection of certain types of people to perpetuate their qualities.

Euthenics. Improving human beings through changes in the environment.

Evolution. Gradual change from one form to a new, or different, form.

Fetus. The developing organism of a human being prior to birth.

Genes. The units in chromosomes that determine hereditary traits.

Genetic condition. Any quality or trait that an individual inherits from his or her parents.

Genetics. The scientific study of heredity.

Genetic code. The process of chemical change and recombination that occurs during cell division, involving the four basic chemical compounds of DNA and the 20 amino acids that make up all proteins.

Genetic trait. Trait determined by genes, not necessarily congenital.

Heredity. The inheritance of physical and mental characteristics.

Martyrdom. Great suffering, often deliberately chosen.

Meiosis. The special type of cell division occurring in the germ cells.

Mitosis. The reproduction of a cell by dividing into an exact duplicate of itself.

Multifactorial. Determined by multiple factors, genetic and non-genetic, yet in a discernible hereditary pattern.

Mutation. A failure of a gene to produce an exact self-copy, resulting in modification of the hereditary trait produced by that gene.

Nutrition. Any form of nourishment, mainly food.

Nucleus. The central part of a cell.

Prenatal care. Maintenance of a mother's health prior to the birth of her child.

Prenatal diagnosis. Determination of the likelihood of birth defects in an unborn fetus.

Psychomotor. Of or relating to motor action directly proceeding from mental activity.

Recessiveness. The quality of a particular trait that does not appear when paired genetically with a dominant trait.

Sex chromosomes. Chromosomes responsible for sex determination.

Sibling. A brother or sister.

Species. Groups of living organisms that interbreed.

Sperm. Male germ (sex) cells that unite with female ova (egg) cells to produce a new organism.

Common Congenital Diseases and Disorders

A Summary of Important Features

Cleft lip/palate

Symptoms: Immediately observable at birth. Cleft lip: failure of the two sides of the upper lip to grow together properly. Cleft palate: a split or opening in the roof of the mouth leading to complications in breathing, speech, hearing, and ingestion of food. These conditions occur both together and separately.

Treatment: Corrective surgery, speech therapy.

Pattern of transmission: Variable, often multifactorial.

Club foot

Symptoms: Twisted position of one or both of a baby's feet, easily recognizable at birth, resulting, if untreated, in inability to walk and/or shortened legs or toes.

Treatment: Surgery, corrective shoes or braces. With proper care, most affected individuals can walk normally by the time they reach physical maturity.

Pattern of transmission: Multifactorial.

Cystic fibrosis

Symptoms: Unusually thick mucus blocks the lungs, causing coughing, difficult breathing, infections, and distended lungs. Secretion of the digestive juices is reduced, causing poor digestion of food, a massive appetite, thin body build, poor tolerance of exercise, short stature, and in some cases delayed sexual maturation. Salt is lost in perspiration more easily than normally.

Treatment: Life expectancy is shorter than normal. Physical therapy can improve breathing; synthetic digestive enzymes can improve digestion; salt tablets can help to avoid loss of salt in perspiration; antibiotics can treat lung infections.

Pattern of transmission: Recessive.

Diabetes

Symptoms: Thirst, increased appetite, weakness, weight loss; in extreme circumstances, unconsciousness or convulsions.

Treatment: Since diabetes is essentially the result of metabolic disorders leading to high blood sugar, this condition can be corrected by insulin injections and careful diet and exercise.

Pattern of transmission: Multifactorial.

Down's syndrome

Symptoms: In early stages of development, distinct physical features: slanting eyes (Down's syndrome is sometimes referred to as "mongolism"), curving folds of skin at the eyes, shorter than average stature, often a single large crease on the palm of the hand. In later stages of development, varying degrees of mental retardation, occasionally heart disease and other complications.

Treatment: Antibiotics for some complications attending Down's syndrome; special education. Life expectancy may be nearly normal.

Pattern of transmission: Chromosome abnormality.

Hemophilia

Symptoms: Poor clotting of blood, spontaneous bleeding or excessive bleeding after minor injury; damage to joints.

Treatment: Injections of the deficient clotting factor. With proper care, hemophiliacs can lead normal lives, although they must take care in exercising.

Pattern of transmission: X-linked.

Huntington's chorea

Symptoms: Between the ages of 30 and 40, progressive deterioration of the brain and central nervous system, producing involuntary jerking, loss of mental abilities, depression, insanity, and ultimately death.

Treatment: None.

Pattern of transmission: Dominant.

Hydrocephalus

Symptoms: Larger than normal head size. Hydrocephalus is the result of abnormal quantities of cerebrospinal fluid in the brain, usually due to a blockage, tumor, or malformation that interferes with the circulation of the fluid through the central nervous system.

Treatment: Surgical removal of excess fluid. Without treatment, affected children rarely survive.

Pattern of transmission: Multifactorial.

Muscular dystrophy

Symptoms: Muscular dystrophy is actually a group of disorders, all of which involve damage to the muscles supporting the skeleton, resulting in progressive weakness. Duchenne-type muscular dystrophy, occurring in the first few years of life, is the most common.

Treatment: Temporary relief through therapy and braces. Death usually occurs within 15 or 20 years of onset.

Pattern of transmission: Commonly X-linked.

Phenylketonuria (PKU)

Symptoms: An inborn error of metabolism, PKU is the inability to metabolize the amino acid phenylalanine. In the newborn PKU can be detected through a simple

blood test. Later on a child with PKU can be identified by unusually lighter hair or skin than his or her siblings. In advanced stages PKU can produce abnormal destructive behavior and degrees of mental retardation.

Treatment: The most serious effects of PKU can be prevented in infancy through a special diet that balances the body's lack of the phenylalanine enzyme. If PKU advances beyond this stage, institutionalization may be required to deal with severe mental and physical retardation.

Pattern of transmission: Recessive.

Rh incompatibility

Symptoms: In the newborn child, jaundice, anemia, stillbirth, or complications leading to mental retardation and subnormal physical development.

Treatment: Rh disease is the result of the incompatibility of Rh blood factors in the mother and father. Although once among the most common causes of birth defects, the disease can now be prevented completely by a vaccine developed in 1968.

Pattern of transmission: Dominant. Rh factor disease can occur only in situations where the father has Rh positive blood while the mother has Rh negative blood.

Sickle cell anemia

Symptoms: A blood test will reveal that the bearer of this disease has blood cells that are sickle-shaped, rather than the normal round shape. Although victims of this disease can lead normal lives, severe anemia is common, and affected individuals experience periodic pain and infections. The life span is usually shorter than normal.

Treatment: No cure has been found. Temporary analgesic relief, antibiotics, and occasional blood transfusions are necessary.

Pattern of transmission: Recessive.

Spina bifida

Symptoms: A defect in the bone structure of the spinal column, often producing a large cyst containing parts of the spinal cord, observable at birth.

Treatment: Corrective surgery. In severe cases, children who have been treated are paralyzed below the waist.

Pattern of transmission: Multifactorial.

Tay-Sachs disease

Symptoms: An infant with Tay-Sachs disease appears normal at birth. Within four to eight months the first symptoms appear in the form of weakness, sluggishness, and poor psychomotor development. The symptoms become progressively more severe—blindness, deafness, seizures, paralysis, and total mental retardation usually occur. Death always occurs by three to five years of age.

Treatment: None.

Pattern of transmission: Recessive.

Thalassemia (Cooley's anemia)

Symptoms: Severe anemia, the result of a failure of the body to produce blood cells with the normal amount of hemoglobin. Children with thalassemia are pale and listless.

Treatment: Hemoglobin transfusions throughout life, beginning in the first year.

Pattern of transmission: Recessive.

Selected Readings

Books

Apgar, Virginia, and Beck, Joan. *Is My Baby All Right?* New York: Trident Press, 1972.

The most complete book yet published on the subject of birth defects, including an account of the process of heredity written in lay terms.

Darwin, Charles. *The Origin of Species.* Numerous editions.

The classic on evolution.

Dobzhansky, Theodosius. *Heredity and the Nature of Man.* New York: Signet Science Library, 1964.

A brief history of genetics and speculations about the future. Comprehensive wisdom clearly written.

Dubos, Rene. *So Human an Animal.* New York: Charles Scribner's Sons, 1968.

This 1969 Pulitzer prizewinner emphasizes the danger of man's losing his humanness to his mechanized surroundings.

Etzioni, Amitai. *Genetic Fix: The Next Technological Revolution.* New York: Harper and Row, 1975.

The morality of the new genetics explored in a chatty, self–centered way as Etzioni recalls an international conference on the subject. Highly readable.

Goldstein, Philip. *Genetics Is Easy.* New York: Viking Compass Books, 1967.

Terse descriptions of Mendel's discoveries.

Halacy, D. S., Jr. *Genetic Revolution: Shaping Life for Tomorrow.* New York: Harper and Row, 1974.

An examination of the moral and ethical issues raised by the new genetics.

McCusick, V. A. *Mendelian Inheritance in Man.* baltimore: Johns Hopkins Press, 1971.

An authoritative catalog of genetic diseases and disorders.

Montague, Ashley. *Human Heredity.* New York: World Publishing Company, 1959.

A well-written argument for deemphasizing the role of genetics and recognizing the importance of environmental factors.

Moore, Ruth. *Evolution.* New York: Time, Inc., 1962.

Loaded with beautiful pictures and lucid diagrams.

Nyhan, William L., M.D. *The Heredity Factor: Genes, Chromosomes, and You.* New York, Grosset & Dunlap, 1976.

An excellent, well written general guide.

Stern, Curt. *Human Genetics.* San Francisco: W. H. Freeman & Co., 1960.

Detailed, scholarly, complete, and richly illustrated.

Watson, James D. *The Double Helix: A Personal Account of the Discovery of the Structure of DNA.* New York: New American Library, Mentor Books, 1968.

A witty, entertaining account of one of the major biological discoveries.

Magazine Articles

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Ausubel, F., "The Politics of Genetic Engineering: Who Decides Who's Defective?" *Psychology Today,* June, 1974, pp. 30ff.

Bylinsky, G., "What Science Can Do About Hereditary Diseases," *Fortune,* September, 1974, pp. 148ff.

Culliton, B. J., "Genetic Screening: The States May Be Writing the Wrong Kinds of Laws," *Science,* March 5, 1976, pp. 926–929.

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Gwynne, P., "Genetic Watchdogs: NIH Guidelines for Recombinant DNA Research," *Newsweek*, July 4, 1976, pp. 106–107.

_____, "Politics and Genes," *Newsweek*, January 12, 1976, pp. 50–52.

Heiserman, D. L., "New Hope in the War on Genetic Disease," *Science Digest*, February, 1973, pp. 68–72.

Judson, H. F., "Fearful of Science," *Harper's*, March, 1975, pp. [illegible]ff.

Lamberg, L., "Genetic Screening: Learning What You Never Wanted to Know," *Today's Health*, March, 1976, pp. 28–31.

Motulsky, A. G., "Brave New World? Ethical Issues Raised by Current Approaches to Prevention, Treatment, and Research on Genetic Diseases," *Science*, August 23, 1974, pp. 653–664.

National Geographic, September, 1976, issue on "The New Biology."

Pines, Maya, "Genetic Profiles Will Put Our Health in Our Own Hands," *Smithsonian*, July, 1976, pp. 86–91.

Restak, R., "The Danger of Knowing Too Much," *Psychology Today*, January, 1976, pp. 64ff.

Time, "Genetic Moratorium: Demands of the Cambridge, Massachusetts, City Council," July 19, 1976, p. 67.

Wade, N., "Recombinant DNA: Guidelines Debated at Public Hearing," *Science*, February 27, 1976, pp. 28–31.

Weinberg, J. H., "Decision at Asilomar," *Science News*, March 22, 1975, pp. 194–196, (decisions about genetic research guidelines).

Periodicals

New England Journal of Medicine

Science

Science News

Available in many libraries.

Instructional Materials for Classroom Use

All About Healthy Me. A workbook, coloring book, and reader for kindergarten through grade three, emphasizing the individual's unique qualities and differences from others.

Available from:
Office of Child Development
U.S. Department of Health, Education, and Welfare
Washington, D.C. 20201

Teacher's manual also available.

So I Have Sickle Cell Trait . . . Booklet for the primary or intermediate level, using smiling-faced characters to tell about sickle cell trait.

Available from:
National Sickle Cell Disease Program
Bethesda, MD 20013

Tony's Sickle Cell Crisis. Fully illustrated coloring and storybooks for the elementary level.

Available from:
Rapid Press
1726 North First Street
Milwaukee, WI 53212

What Are the Facts About Genetic Disease? Booklet. A brief introduction with informative charts and statistics. Appropriate for intermediate and secondary levels.

Available from:
National Institutes of Health
Bethesda, MD 20014

Why Is Mary on a Diet? Illustrated reader for intermediate level about a little girl who was born with PKU and must be on a special diet for most of her life.

Available from:
The Children's Memorial Hospital
2300 Children's Plaza
Chicago, IL 60614

Instructions on how to perform genetic experiments with fruit flies, corn seedlings, etc., are available with the purchase of materials for experiments from several companies.

Ward's Natural Science
P.O. Box 1749
Monterey, CA 93940

Nasco Biological Science
P.O. Box 3837
Modesto, CA 95352

College Biological Supply Co.
P.O. Box 1326
Escondido, CA 92025

Agencies

For further information about additional booklets, films, filmstrips, learning materials, or speakers, contact:

The Arthritis Foundation
1212 Avenue of the Americas
New York, NY 10036

California Epilepsy Society
14652 Ventura Boulevard
Sherman Oaks, CA 91403

National Hemophilia Foundation
25 West 39th Street
New York, NY 10006

Muscular Dystrophy Associations of America, Inc.
810 Seventh Avenue
New York, NY 10019

National Association for Retarded Citizens
P.O. Box 6109
Arlington, TX 76011

National Cystic Fibrosis Foundation
3379 Peachtree Road, NE
Atlanta, GA 30326

National Foundation/March of Dimes
P.O. Box 2000
White Plains, NY 10602

National Genetics Foundation
250 West 57th Street
New York, NY 10019

Sickle Cell Anemia Research and Education, Inc.
2201 Steiner Street
San Francisco, CA 94115

State of California Regional Centers for the
Developmentally Disabled
Regional Centers Section
744 P Street
Sacramento, CA 95814

Questionnaire
for Pretest or Review

Recommended for teachers, secondary students, and adult education students, as either a pretest or review of instructional content.

Instructions. For each true/false and multiple choice question below, circle one or more answers that apply.

1. A person can acquire a genetic disease from:
   A. Poor diet
   B. Inheritance
   C. Friends
   D. Environment

2. Some diseases are passed on from parent to child through heredity.
   A. True
   B. False

3. Which of the following are inherited?
   A. Sickle cell anemia
   B. Hemophilia
   C. Diabetes
   D. Tay-Sachs disease
   E. Cystic fibrosis
   F. Tuberculosis
   G. Pneumonia
   H. Chicken pox

4. A gene is:
   A. The smallest unit of heredity
   B. A part of the red blood cell
   C. An aid to indigestion

5. A carrier of a genetic disease has the following characteristics:
   A. Must have a blood tranfusion to correct his or her condition
   B. May be unaware of his or her carrier state
   C. Will never suffer from the disease itself
   D. Has a short life expectancy

6. Recessive genetic diseases and disorders:
   A. May come and go
   B. Are inherited from both parents
   C. May be easily recognized in the carrier

7. Dominant genes:
   A. Can be easily recognized in the affected individual
   B. Bring out the recessive genes
   C. Are not important in the occurrence of genetic diseases and disorders

8. Match the racial groups which are most likely to have the indicated conditions:

A. Sickle cell anemia	(1) Blacks
B. Diabetes	(2) Caucasians
C. Cystic fibrosis	(3) Jews
D. Tay-Sachs disease	(4) Mediterraneans
E. Down's syndrome	(5) Native Americans
F. Thalassemia	(6) Chicanos
	(7) All of the above

9. Cystic fibrosis causes:
   A. Lung problems
   B. Obesity
   C. Digestive problems
   D. Ringing in the ears
   E. All of these
   F. None of these

10. Sickle cell anemia is a recessive genetic disorder.
    A. True
    B. False

11. A person who has diabetes:
    A. Needs to have regular meals, snacks, and exercise
    B. Should stay indoors at all times
    C. Needs to eat extra sugar with meals

12. Tay-Sachs disease cannot be diagnosed before the teenage years.
    A. True
    B. False

13. To obtain information about genetic disease, which of the following resources could be used?
    A. Local school board
    B. Welfare department
    C. National Cystic Fibrosis Foundation
    D. National Foundation/March of Dimes
    E. Local library

14. Genetic diseases:
    A. Are the subject of increasing amounts of research
    B. Can cause great economic stress on affected families
    C. Can influence the development of a positive self-image of an affected child

15. Genetic engineering is a new field of study and can be both a promise and a threat for the future.
    A. True
    B. False

Answers on next page

Answers

1. B
2. A
3. A, B, C, D, E
4. A
5. B, C
6. B
7. A
8. A. Sickle cell anemia — (1) Blacks
   B. Diabetes — (7) All of the above
   C. Cystic fibrosis — (2) Caucasians
   D. Tay-Sachs disease — (3) Jews
   E. Down's syndrome — (7) All of the above
   F. Thalassemia — (4) Mediterraneans
9. A, C
10. A
11. A
12. B
13. C, D, E
14. A, B
15. A

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